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**ADVANCING SCIENCE AND CONTROLLING THE MISUSE OF GENETIC
INFORMATION IN EMPLOYMENT AND INSURANCE – TOWARDS AN
EFFECTIVE EUROPEAN UNION REGULATORY FRAMEWORK**

This thesis is submitted to the National University of Ireland, Galway in fulfilment of
the requirement for the degree of

Doctor of Philosophy

By

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Abstract

The science of genetic testing and related technology is rapidly advancing. Among other things, genetic testing technology may offer the prospect of being able to detect the onset of future disabilities. It also offers the possibility of personalised medicine, which potentially promises to usher in a health care revolution, with significant public health benefits. It has the potential to transform health care through earlier diagnosis, more effective prevention and treatment of disease, and avoidance of drug side effects. Genetic technology is becoming more advanced and sophisticated and is being used increasingly for both medical and non- medical purposes.

If sufficient protections are not in place to prohibit the misuse or discriminatory use of genetic information by third parties (for example, employers and insurance companies) then such a legislative and policy vacuum could further setback the inclusion of persons with putative disabilities (and older persons) in the community. In addition, such a legislative and policy vacuum may also negatively impact upon public confidence in genetic science and technology, which may hamper public health and economic growth. To date, there is no European Union level regulation protecting the privacy of genetic information or protecting against the discriminatory use of such information.

Against the backdrop of rapidly advancing genetic technologies and the ethical and legal concerns that arise, this thesis examines the need for an EU level regulatory framework. The main focus of this examination is on the need to control the misuse of genetic information in the areas of employment and insurance, with the objective of advancing genetic science and acknowledging the competing interests at stake. This thesis will also consider the appropriate shape and content of a potential EU level regulatory response.

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Additional Protocol to the Convention on Human Rights and Biomedicine concerning genetic testing for health purposes (2008)

Council of Europe Recommendations

Recommendation No. R (90) 13 of the Committee of Ministers to Member States on Prenatal Genetic Screening, Prenatal Genetic Diagnosis and Associated Genetic Counselling (Adopted by the Committee of Ministers on 21 June 1990 at the 442nd meeting of the Minister's Deputies)

Recommendation No. R (92) 3 of the Committee of Ministers to Member States on Genetic Testing and Screening for Health Care Purposes (Adopted by the Committee of Ministers on 10 February 1992 at the 470th meeting of the Minister's Deputies)

Recommendation No. R (97) 5 on the Protection of Medical Data (Adopted on 13 February 1997)

Recommendation CM (Rec 2010) 11 of the Committee of Ministers to Member States on impact of genetics and training of health organisation of health care services and training of health professionals (Adopted by the Committee of Ministers on 29th September 2010 at the 1094th meeting of the Minister's Deputies)

List of Abbreviations

ALRC	Australian Law Reform Commission
CFREU	Charter of Fundamental Rights of the European Union
CoE	Council of Europe
CRPD	United Nations Convention on the Rights of Persons with Disabilities
ECHR	European Convention on Human Rights
ECtHR	European Court of Human Rights
EEOC	Equal Employment Equality Commission
EIOPA	European Insurance and Occupational Pensions Authority
ESC	European Social Charter
EU	European Union
FRA	Fundamental Rights Agency of the European Union
GINA	Genetic Information Non-Discrimination Act
HGP	Human Genome Project
TEU	Treaty on the European Union
TFEU	Treaty on the Functioning of the European Union
US	United States
UK	United Kingdom
UN	United Nations
UNESCO	United Nations Education
UDHGHR	Universal Declaration on the Human Genome and Human Rights
IDHGD	International Declaration on Human Genetic Data
UDBHR	Universal Declaration on Bioethics and Human Rights

Chapter 1: Introduction: Aims, methodology and structure

1. Introduction

This chapter will introduce the thesis, and the research question proposed. It will detail the aims and scope of the thesis. This introduction will also highlight the main methodologies used throughout this thesis. The final section of this chapter will set out a detailed outline of the four parts to this thesis and the content of the individual chapters.

1.1 Background to research

Unprecedented scientific and technological advances have led to an increase in the practice of genetic testing for medical and non-medical purposes. Genetic science and technology is advancing at a rapid pace, with scientists continuing to make genetic discoveries as to the make up of the human body and the cause and effect of disease and disability. These discoveries are facilitating technological innovation and the development of a range of genetic tests that are becoming more sophisticated. Therefore, as genetic science continues to advance, genetic technologies inevitably become more accessible and available.

Among other things, genetic testing technology offers the prospect of being able to detect the onset of future disabilities. Therefore, advancing genetic science, in conjunction with an ageing population is resulting in more and more individuals being exposed as having putative disabilities. The increasing availability of genetic information, together with the insights it offers into an individual's future health and predisposition to disability is valuable information to a wide range of third parties (including employers), as well as providers of social goods and services (such as insurers).

In light of the potential for misuse of this information, the need arises to control the flow and use of genetic information, and the question falls to be considered whether an adequate regulatory framework exists to address these ethical and legal issues and to protect an individual's fundamental human rights. It is also necessary to address the evident conflict of competing legitimate rights that arise. In the absence of an appropriate regulatory framework, it is anticipated that more and more individuals will be excluded from a range of social and economic goods and services, potentially leading to the creation of a genetic underclass. The absence of an appropriate regulatory framework also has the potential to stifle scientific and technological progression, with adverse implications for both public health and economic growth.

1.2 Research questions and scope of research

Against the backdrop of rapid scientific and technological developments, the main question in this thesis is to address the need for and the options available for a EU level regulatory response to address and control the use of genetic information by third parties, namely employers and insurance companies. On consideration of an EU level response, this thesis aims to elucidate the key ethical and legal issues arising and acknowledge the legitimacy of the competing interests at stake. Specifically it will address the discriminatory use of genetic information and the privacy implications of disclosing genetic information, as well as the wider societal and public policy implications of misusing genetic information. This research question is considered in light of increasing advances in genetic science and technology and in consideration of the range of fundamental human rights at stake, particularly as regards putative persons with disabilities. There are also a myriad of commercial interests at stake, which are likely to become more amplified as science and technology advance. In addition, this thesis will address whether a legislative response alone is sufficient to address this area. In this regard, it considers the potential need for an awareness raising campaign and a human genetics advisory network.

This thesis takes a general human rights based approach to this area. It acknowledges the protection of genetic information as a human right, which merits recognition. This is in line with the recognition of disability as a human rights issue, which is slowly becoming an accepted norm globally. Specifically, this thesis therefore takes a disability rights approach to this area. This approach recognises the connection between genetics and disability, which is expressed in this thesis primarily through the theory of the social model of disability and its application in this area.

It is acknowledged that although genetic testing by third parties has not yet been documented as taking place on a widespread basis, and notwithstanding the fact that misuse of genetic information is evidently not yet a serious problem, rapidly advancing genetic technology indicates that testing will undoubtedly become cheaper, more sophisticated and therefore more accessible to third parties. This thesis recognises the need to respond preemptively to these issues at an early stage, to anticipate the potential orientation of the technological advances and respond accordingly.

This thesis takes an interdisciplinary approach to this area. It acknowledges the need to be aware of the reality of advancing genetic science and technology. It provides a scientific perspective to this discussion and appreciates the need to ensure that science and technology progress. Accordingly, any new regulatory response has to be calibrated with the need not to inhibit the advancement of science and the development of new genetic testing technologies. The consideration of an appropriate regulatory framework at EU level therefore also proves crucial in maintaining public confidence in science without which it cannot prosper for the benefit of all individuals and for public health, as will be explored.

Although this thesis is narrowly focused, it acknowledges that there are wider ethical, legal and social issues that arise at the forefront of rapid scientific advances. For example, from an ethical and philosophical perspective, such advances may change the concept of humans and what it means to be human. Science may take over in conceptualising human beings, which may force society to construct a new language to frame human rights. The issues addressed in this thesis are therefore part of a wider web of 21st century challenges arising at the interface of science, ethics and the law.

In considering the case for EU level action in this area, this thesis conducts a comprehensive analysis of the current regulatory landscape, at an international level, EU level, and in the relevant comparative jurisdictions. From a regulatory perspective, this thesis examines the non- discrimination, privacy (data protection) and property theories. The reasons for considering these regulatory frameworks are outlined in chapter 5. It is noted that this is a legal doctrinal thesis. It relies upon existing theories of regulation including non- discrimination, privacy and property. It applies these existing theories to the area of genetic technologies and genetic information. Accordingly, this thesis does not propose any new regulatory theories in this area.

This thesis identifies the appropriate legal framework, as well as the moral impulse for action in this area, particularly at international level. It identifies the gaps in protection, the evident patchwork of protection amongst the EU Member States and the compelling reasons for protection of genetic information. On proposing a case for EU level action, this thesis recommends the consideration of the first genetic information non- discrimination directive. Consideration of such a potential legislative framework necessitates an examination of the EU's competence to act to address discrimination on the grounds of disability, as well as the EU's competence to act to ensure the functioning of the EU internal economic market.

This thesis presents a comprehensive, thoroughly researched and informed analysis of the ethical and legal dilemmas arising at the frontiers of genetic science in light of the current international, comparative and European framework. In accordance with main research question, the key objective is to identify the gaps in protection and consider the need for the first genetic information non- discrimination directive at EU level.

2. Methodology

This section will give a brief overview of the methodologies used in this thesis.

2.1 Advanced legal methods and research

The main methodological tool utilised in this thesis is advanced legal methods and research. This involved undertaking a significant amount of library based research. It also involved indepth and detailed searches of all relevant legal databases. This research allowed me to engage in traditional legal analysis of a vast amount of primary and secondary sources, including statutory material, academic commentary, and materials such as policy reports and recommendations, and Law Reform Commission reports from various jurisdictions. It also included analysis of academic textbooks and journal articles. This research and analysis allowed me to recognise the key themes and issues arising. It also facilitated identification of the gaps, assisting me to refine and focus my thesis on the key issues. This methodology was used in all of the chapters.

In ascertaining the question of evidence of genetic and other misuse of genetic information, this thesis examines existing empirical evidence in this area. This thesis does not engage in any additional empirical research.

2.2 Interdisciplinary research

This thesis also engages in interdisciplinary research as a methodological tool. This necessitated an analysis of a variety of different fields. In this regard, the research commenced with a detailed, comprehensive review of the science of genetics, the concept of genetic testing and other genetic technologies, to provide an understanding and discussion of the area of genetic science. It involved analysis of scientific literature and other materials, using traditional doctrinal analysis methods.

The analysis of genetics also involved a series of meetings and discussions with leading experts in the area of genetic science at NUI Galway. Meetings were organised with a number of these experts to learn more about the basic elements of and the history of genetic science, as well as the future of advancing genetic technologies. This interdisciplinary approach also necessitated a clear understanding of ethics and the particular ethical issues arising in the area of genetic science. This research involved analysis of legal materials, philosophical materials and sociological materials. This involved traditional doctrinal analysis of various sources.

This interdisciplinary approach was also necessary in light of the disability approach taken in this thesis. This research involved traditional doctrinal analysis of the different models of disability, with a particular focus on the social model of disability theory. It necessitated examination of both legal sources (primary and secondary sources), as well as other relevant philosophical and sociological materials. This analysis contributed towards shaping the understanding of genetic discrimination in this context and identifying connections between genetics and disability.

2.3 Comparative analysis

Another methodological tool used in this thesis is comparative analysis. This involved examining the legal position in several jurisdictions such as the United States (US), Australia and the EU. This involved a considerable amount of research by looking at the position in these various jurisdictions to determine the effectiveness of policy and legislative models in governing the issue of genetic discrimination.

In order to carry out this comparative study, it was necessary to understand the legal framework and background within which the various laws were implemented, as well as the different policies and reasons behind the laws. This involved traditional doctrinal analysis of literature and academic commentary, as well as analysis of various secondary sources and materials. Research was carried out on the numerous recommendations, policy reports, and congressional hearings. This part of the the thesis involved analysis of the various legislative instruments and other regulatory mechanisms used such as moratoria.

This comparative element also included an overview of the international human rights position, which examined in particular, the relevant United Nations (UN) treaties and soft law instruments. In addition, the research conducted in this section involved consultation with various individuals and organisations at national, European and international level, for example, the Irish Data Protection Commissioner, the European Disability Forum, the European Society of Human Genetics, and the American Society of Human Genetics.

2.4 Field trips

The research carried out in this thesis also involved a number of field trips. The first research trip was to Washington DC in April 2011. During this time I met with leading US experts at the American Society of Human Genetics¹ and Genetics Alliance,² to learn more about the legislative endeavours in the US. Another research trip was to the Burton Blatt Institute, Syracuse University, New York, in October 2012 under the guidance of Professor Peter Blanck, (Chairman of the Burton Blatt Institute). During this time I met with leading experts in the area of disability law and policy and bioethics. I also gave a number of presentations on my research and theories proposed during these field trips.

In addition to the above field trips, I have also attended and participated in many key conferences in the area, including the European Society of Human Genetics Annual Meeting (2012) and the American Society of Human Genetics Annual Meeting (2011 and 2012).

¹ <http://www.ashg.org/> (accessed 5 August 2013)

² <http://www.geneticalliance.org/> (accessed 5 August 2013)

2.5 Other research methods

In 2011 and 2012 I organised two conferences which explored the regulation of genetic information at EU level, and which took place at NUI Galway and the European Parliament. These conferences proved key to shaping the theories used, as well as the structure of the thesis. These events highlighted the interdisciplinary approach taken throughout this thesis and they focused on the interaction between genetic science, ethics and the law, and how best to regulate this area.

The research carried out in this thesis involved consultation with various individuals and experts (at national, European and international levels) in the scientific, sociological, legal and policy fields. This element of research was used throughout the thesis.

Over the past four years, I have also carried out a number of key informant interviews with leading experts in the areas of international law, disability law and bioethics. Interviews were held with, *inter alia*, Peter Blanck (Burton Blatt Institute, Syracuse University), Anita Silvers (Department of Philosophy, San Francisco State University), Michael Stein (Harvard Law School Project on Disability), Janet Lord (Burton Blatt Institute, Syracuse University, formerly of Blue Law International), Yann Joly (Centre of Genomics and Policy, McGill University), Joann Boughman (American Society of Human Genetics) and Ine Van Hoyweghen (University of Leuven, formerly of Maastricht University).

3. Structure of thesis

This section will give an overview of the structure of this thesis and outline the content of the different sections and chapters. It comprises of four parts.

Part 1, which is entitled 'New science and technology and the ethical and legal issues arising', includes three chapters. Chapter two details the history of genetic science and the future of advancing genetic technology. The objective of this chapter is to illustrate the reality and speed at which genetic science and technological innovation is advancing. It will also examine the limitations of genetic science and technology, with particular reference to the predictive value of genetic technologies. This chapter aims to highlight how advancing science and technology raises a myriad of ethical and legal issues with the resulting deluge of genetic information that is increasingly becoming available to individuals and third parties.

Chapter three looks at the general ethical and legal issues and the public policy concerns arising from advancing genetic science and technology. It is noted that the ethical and socio- political assumptions that underlie the thesis are assumed on the basis of already existing literature, for example, as is set out chapter

three. This chapter explores the ethical and legal issues arising, from different perspectives, such as the individual, family members and third parties. It also looks at the broader societal and public policy concerns that arise in respect of the use and misuse of genetic science. The main aim of this chapter is to demonstrate the tension that exists between the various competing rights and to provide some context for the ethical aspect of this discussion. Although it is acknowledged that misuse of genetic information can take place in a range of third party contexts, it narrows the focus to employment and insurance, in light of the fact that employment and insurance are both gateways to accessing a variety of social and economic goods and services.

Chapter four highlights the disability perspective taken in this thesis. It examines the relevance of the social model of disability and its application in this discussion. It is acknowledged that abuse of genetic technologies in these contexts exacerbates the susceptibilities of persons with putative disabilities, who are already in a vulnerable position in society and who already may be excluded. Against a disability rights framework, the main aim of this chapter is to highlight the current best thinking on the social construct of disability and how it can shape the discussion and regulation in this area.

Part 2 is entitled 'The imperative for regulation and the choice of regulatory frameworks' and it includes three chapters. Chapter 5 explores the mode of regulation and the choice of regulatory frameworks in legal theory. It examines the non-discrimination approach, the privacy approach and the property approach. It evaluates these regulatory frameworks, by exploring the merits and shortcomings of each, with a view to ascertaining the most appropriate model to effectively control the use of genetic information.

Chapter 6 gives an overview of the international human rights position. This chapter will examine UN treaty law and jurisprudence. In light of the disability approach taken in this thesis, this will focus particularly on the United Nations Convention on the Rights of Persons with Disabilities (CRPD) and its potential to shape our interpretation of the issues, and provide an international forum for discussion. This chapter will also examine UN soft law instruments, which illustrate the growing concern for the protection of genetic information at international level and indicate a need for regulatory interventions in this area. The human rights framework of the UN provides a moral imperative for action in this area. Chapter 7 is entitled 'Evolving comparative benchmarks'. This chapter examines the position in the US and Australia. The aforementioned countries were selected, as these are two of the most developed jurisdictions, from a legislative and policy perspective. This chapter analyses how these jurisdictions have responded to the issue of advancing genetic science and technology and how the ethical and legal issues have been addressed. It also highlights the choice of regulatory framework adopted in the respective jurisdictions.

Part 3 is entitled 'The European perspective' and it includes two chapters. Chapter 8 is entitled 'Human rights norms in the Council of Europe and the regulation of genetic information.' This chapter provides a chronological overview of the relevant instruments that shape the Council of Europe's human rights framework (including the European Convention on Human Rights and the Council of Europe Convention on Human Rights and Biomedicine). It will also highlight other relevant policy instruments and recent developments, which shape the Council of Europe's position in this area, and will highlight the evolution and interpretation of human rights. The analysis of these instruments will also illustrate the position of the Council of Europe in response to emerging genetic technology, in providing a moral and ethical framework from which to approach this area.

Chapter 9 is entitled 'The EU and genetic information.' This chapter looks at human rights law in the EU, with a focus on the Charter of Fundamental Rights of the EU (CFREU). This chapter looks at the scope of the current EU position in the area of data protection and non-discrimination, including recent developments in these areas. It also highlights other relevant policy perspectives in the EU in respect of genetic information. The main objective of this chapter is to explore the extent to which current EU law applies to and protects genetic information, and to identify the gaps in protection.

Part 4 is entitled 'Building the case for a EU regulatory framework' and includes two chapters. Chapter 10 builds a case for EU action. It firstly presents an argument for EU action from the current diversity of approaches. This includes an overview of the position in some of the individual EU Member States, illustrating the variety of approaches taken and the evident patchwork of protection. In building the case for EU level action, chapter ten will also argue that such a legislative framework is necessary in order to enhance public trust and confidence in genetic science and technology. This is a key argument from the perspective of public health, as well as EU innovation and economic growth.

In proposing EU level action, it will also provide an overview of the evidence of misuse of genetic information. In terms of EU competence to act, it discusses the potential legal basis for action at EU level. It will also look at the potential legislative options, and what shape such a response might take. This chapter will also examine the challenges that might arise in considering EU level action. In addition, this chapter will consider whether a legislative response alone is sufficient to address this area. Chapter 11 is a final chapter containing conclusions and recommendations. It will connect the various sections of the thesis and make final recommendations as to the regulatory choice and the shape of the law.

PART 1: New Science and Technology and the Ethical and Legal Issues arising

Part 1 of this thesis will firstly provide a clear scientific framework from which to view and contextualise the issues arising in this area. It will chronicle the history of genetic science and highlight the potential for advancing genetic technologies to enhance health care, by offering insights into genetic profiles. It also offers the potential of detecting potential future disabilities which is key to this discussion. This part will highlight the increasing accessibility to and availability of genetic information, for medical and non- medical purposes. Part 1 will then explore the ethical and legal issues arising, as well as some of the broader public policy concerns which shape this debate. In the third party context, it will focus on the employment and insurance fields and highlight the conflict of competing rights that arises. Finally, part 1 will highlight the disability approach taken in this thesis. Part 1 will highlight the potential for misuse of genetic information, the fundamental human rights at stake and the need for a clear regulatory framework to address this area.

Chapter 2: The Background: Advancing Genetic Science

1. Introduction

Genetics is the science of inheritance, and the study of how particular traits and characteristics are passed on from parents to offspring, from physical traits such as height or eye colour to complex disease susceptibilities such as heart disease and different types of cancer.¹ It focuses on genes and their effects on disease and heredity.² Medical genetics is therefore concerned with gaining a greater understanding of disease and the interaction between disease and genes in the human body. In the context of this discussion, it is necessary to provide a conceptual framework of the science of genetics, and formulate a debate that is fully informed of advancing genetic technologies.

The aim of this chapter is to explore the reality (as well as limitations) of advancing genetic science and the rapid pace of technological discoveries and innovation. This raises questions as to the implications of the resulting deluge of genetic information becoming widely available and accessible. This chapter chronicles the evolution of genetic science and technology, including the history of genetics, with a view to providing a basic understanding and insight into the various elements of genetic science. It will outline the endeavours of the Human Genome Project (HGP) as a major international scientific project in this area, and its success in accelerating the pace of genetic science. The HGP was instrumental in the rapid pace of genetic discovery and the development of genetic technologies.

This chapter will also explain the potential of genetic testing to detect the onset of future disabilities, offer the possibility of personalised medicine, and the possible treatment and even cures for diseases at an early stage. Genetic testing and other genetic technologies are becoming more accessible and sophisticated and such technologies are being increasingly used for both medical and non- medical purposes. The increasing availability of genetic information produces a myriad of emerging ethical and legal dilemmas, creating a minefield of problems if not appropriately addressed.

2. The history of genetic science

¹ See generally Burton Guttman *et al*, *Genetics: The Code of Life* (The Rosen Publishing Group, Inc 2011). See also Thomas D. Gelehrter, Francis S. Collins and David Ginsburg, *Principles of Medical Genetics* (2nd edn, Williams & Wilkins 1998)

² Ibid

In tracing the history of genetic science, it is observed that the study of heredity began approximately two hundred years ago.³ At this time, on considering the origins of human beings, Charles Darwin pioneered the concept of natural selection, based on his theory of evolution.⁴ Pursuant to his theory, Darwin postulated that all plants and animals had evolved from a few common ancestors by means of natural selection.⁵ According to Darwin's theory, natural selection dictates which of a species are weakest and therefore die first, and which of the species are stronger, and more likely to live and reproduce.⁶ However, Darwin did not uncover the relevant components of inheritance.⁷ In the mid nineteenth century, Austrian monk Gregor Mendel⁸ endeavoured to uncover the process of inheritance and observed that organisms inherit characteristics through units of inheritance, now referred to as genes.⁹ Mendel engaged in the practice of pea breeding, with a view to learning more about the inheritance patterns in peas.¹⁰ He observed that different factors indicated the different elements of the pea's appearance, for example, the colour and shape of the pea.¹¹

Mendel's insights provided the theoretical basis for dominant and recessive conditions.¹² Mendel proposed that genetic information was passed from parent to offspring through what he deemed "*factors*" (now known as "*genes*").¹³ Mendel's important observation was that the factors occurred in pairs, with one member of the pair being passed on from each parent. He discovered that the two factors which determine a trait may have contradicting instructions, and in these circumstances, it is the dominant aspect which determines appearance.¹⁴ However, pursuant to Mendelian genetics, "*the other factor would persist in latent form, and its effects could reappear in later generations in predictable ratios.*"¹⁵ In 1865 Mendel published a paper entitled 'Experiments in Plant Hybridization' in

³ See generally, Daniel L Hartl and Vitezslav Orel 'What did Gregor Mendel Think he Discovered?' (1992) 131 *Genetics* 245. See also Robin Marantz Henig, *The Monk in the Garden: The Lost and Found Genius of Gregor Mendel, the Father of Genetics* (Mariner Books 2001)

⁴ Charles Darwin, *On the Origin of Species* (John Murray, London 1859). See also, Charles Darwin, *The Descent of Man* (D. Appleton & Co. 1871)

⁵ For further discussion, see Brian Charlesworth and Deborah Charlesworth, 'Anecdotal, Historical and Critical Commentaries on Genetics: Darwin and Genetics' (2009) 183 *Genetics Society of America* 757

⁶ *Ibid*

⁷ *Ibid*

⁸ Arnold Sorsby, 'Gregor Mendel' (1965) 1 *British Medical Journal* 333

⁹ For further discussion, see Brian Charlesworth and Deborah Charlesworth, 'Anecdotal, Historical and Critical Commentaries on Genetics: Darwin and Genetics' (2009) 183 *Genetics Society of America* 757

¹⁰ Thomas Morgan *et al*, *Mechanism of Mendelian Heredity* (Henry Holt and Company 1915) 1

¹¹ *Ibid*

¹² For further discussion, see Garland E. Allen, 'Mendel and modern genetics: the legacy for today' (2003) 27 *Review Endeavour* 2 63

¹³ *Ibid*

¹⁴ *Ibid*

¹⁵ Eric S. Lander and Robert A. Weinberg, 'Pathways of Discovery- Genomics: Journey to the Center of Biology' (2000) 287 *Science* 5459 1777, 1778

which he put forward the principles of inheritance.¹⁶ Mendel's theories were largely ignored at the time, however following his death, his work generated more interest. The concept of Mendelian genetics was rediscovered and is now engrained in the history of genetic science.¹⁷ Indeed, Mendel's theories of inheritance have become a fundamental tool in their application to a variety of organisms, including human beings.

Subsequent important genetic discoveries were made throughout the 20th century. In 1910, Thomas Morgan's research on fruitflies indicated that genes sit on chromosomes, resulting in the theory of the linear arrangement of genes on chromosomes.¹⁸ His work also demonstrated the important insight that random mutations appeared in flies, thereby indicating the theory that random mutations can also occur in human genes.¹⁹ Alfred Sturtevant, who worked with Morgan, was also instrumental in the understanding of chromosomes and used the concept of genetic linkage to illustrate how genes are situated in a linear manner on the chromosome.²⁰ He proposed that genes were linked because they were carried by the same chromosome and in 1913 he presented the first map of a chromosome.²¹

Following initial genetic discoveries in the late 19th century and early 20th century, the modern study of genetics began in 1953 when Crick and Watson discovered the double helix structure of deoxyribonucleic acid (DNA) and published an explanation of the structure of DNA and the manner in which the genes operated.²² This discovery sparked a renewed interest in the science of genetics and the potential to gain a greater understanding of the basis of disease. In 1962, Watson and Crick shared the Nobel Prize in Medicine or Physiology for their discovery of the DNA structure.²³ The discovery of the double helix structure of DNA was a major breakthrough in the genetic revolution in uncovering the structure and working mechanisms of DNA and in the intervening years scientists began uncovering the components of the human genome, and discovering the genetic basis of disease.²⁴ In the 1970s, additional scientific discoveries about

¹⁶ Gregor Mendel, *Experiments in Plant Hybridization* (first published 1865 and 1909, Cosimo Inc. 2008)

¹⁷ Eugene V Koonin, *The Logic of Chance: The Nature and Origin of Biological Evolution* (FT Press Science 2012)

¹⁸ Morgan described his theory of the linear arrangement of genes in the chromosomes in his publication, Thomas Morgan *et al*, *Mechanism of Mendelian Heredity* (Henry Holt and Company 1915)

¹⁹ For further discussion, see John B. S. Haldane, 'The rate of mutation of genes' (1931) 31 *Journal of Genetics* 3 317. See also, John W. Drake, 'Rates of spontaneous mutation' (1998) 148 *Genetics* 4 1667

²⁰ Alfred Henry Sturtevant, *A History of Genetics* (First published in 1965, Cold Spring Harbor Laboratory Press 2001)

²¹ *Ibid*

²² James D. Watson and Francis H.C. Crick, 'Molecular Structure of Nucleic Acids: A Structure for Deoxyribose Nucleic Acid' (1953) *Nature* 737 (announcing the discovery of the DNA double helix).

²³ *Ibid*. See also, James D. Watson 'The Double Helix (a personal account of Watson and Crick's discovery of the structure of DNA)' (Scribner 1998)

²⁴ See generally, Francis Crick, 'The double helix: a personal view' (1974) 248 *Nature* 766

principles of recombinant DNA analysis²⁵ indicated the development of more sophisticated techniques for identifying genes. These discoveries provided the basis and the foundations for the HGP and subsequent genetic developments. Before highlighting the potential of genetic technology, the next section will briefly explore the biology underlying genetic science.

2.1 The biology behind genetic science

In order to provide a more detailed description of the science of genetics, this section will illustrate the mechanisms that contribute towards our understanding of the human genome and the framework within which current and future genetic advances are based. A starting point for exploring genetic science is by explaining the concepts of DNA and genes. Each gene is part of a large molecule of DNA, the structure of which was discovered in 1953.²⁶ The usual model of DNA is in the form of a double helix and resembles a twisted ladder.²⁷ The rungs of the ladder are the base pairs: the union of two of the four molecules, known as nucleotides, each one from an opposing strand of DNA.²⁸ Specifically, this double helix structure is partially composed of four nucleotide bases, guanine (G), cytosine (C), adenine (A), and thymine (T).²⁹ The bases occur in pairs, and the arrangement of these pairs is called the sequence. The sequence of the base pairs in the DNA encodes the genetic information.³⁰ DNA is contained in forty-six thread like chromosomes, numbered according to size, which are found in every cell in the human body.³¹

A gene can be described as *“a piece of DNA, the chemical responsible for storing and transferring all hereditary information in a cell.”*³² Genes achieve this *“by containing recipes for making proteins, described as the true workhorses of all our trillions of cells.”*³³ Therefore, all living organisms consist of proteins, which supply the internal workings of all cells *“as well as specialized enzymes for all essential chemical reactions.”*³⁴ Through these proteins, genes establish bodily structures and functions, as well as how the body operates, for example, the

²⁵ For further discussion, see Allen M. Maxim and Walter Gilbert, ‘A New Method for Sequencing DNA’ (1977) 74 Proceedings of the National Academy of Science 560; Frederick Sanger and A.R. Coulson, ‘A Rapid Method of Determining Sequences by Primed Synthesis with DNA Polymerase’ (1975) 94 Journal of Molecular Biology 3 441

²⁶ James D. Watson and Francis H.C. Crick, ‘Molecular Structure of Nucleic Acids: A Structure for Deoxyribose Nucleic Acid’ (1953) Nature 737

²⁷ Peter B. Dervan, ‘Design of Sequence Specific DNA – Binding Molecules’ (1986) 232 Science, New Series 4749, 464, 464

²⁸ Thomas D. Gelehrter, Francis S. Collins and David Ginsburg, *Principles of Medical Genetics* (2nd edn, Williams & Wilkins 1998) 10- 11

²⁹ Ibid.

³⁰ Robert A. Weinberg, ‘The Dark Side of the Genome’ (1991) 91 Technology Review 3 44, 44

³¹ Ibid

³² Denise K. Casey, ‘What can the new gene tests tell us?’ (1997) 36 The Judges Journal 14, 14

³³ Ibid

³⁴ Ibid at 14

metabolism of food. Genes dictate the production of proteins which, in turn, determine the function of each cell, as well as physical traits, characteristics, predisposition to disease and disability, which are all passed on through the generations.³⁵ Disease generally occurs when mutations, or genetic flaws arise (in conjunction with external influences).³⁶ Genes vary slightly from individual to individual, which, together with external factors such as environmental influences, lifestyle choices and diet, influence the expression of an individual's traits.³⁷

Chromosomes are also a crucial element to understanding the science of inheritance. Genes are grouped together in structures called chromosomes. A chromosome is a very long strand of DNA. Each human cell has forty- six individual chromosomes contained in twenty- three pairs.³⁸ An individual inherits one chromosome from each pair from either parent.³⁹ The complete set of genes for a human being is called the human genome. The human genome comprises the twenty- three pairs of chromosomes that all humans possess and within which all human genes are contained. A genome is effectively the sum of the genetic information that is stored in cells and passed from one generation to the next.⁴⁰ The genome is central because it indicates how cells operate and how organs interact.

Having highlighted the various biological elements which constitute the human genome, this provides a scientific framework within which to further explore the intricacies of genetic science and to illustrate the reality and the potential of advancing technology.

2.2 Genetic conditions and disorders

The discoveries in genetic science made throughout the 20th century contributed towards a greater understanding of the genetic make- up of human beings, as well as our understanding of the connection between disease and genes and the influence of external factors. In order to fully understand the science of genetics and its potential to improve our knowledge of genes and disease, it is necessary to outline the different types of genetic conditions. Together with general characteristics that determine an individual's physical appearance, such as

³⁵ Eric Mills Holmes, 'Solving the Insurance/ Genetic Fair/ Unfair Discrimination Dilemma in Light of the Human Genome Project' (1996- 1997) 85 Kentucky Law Journal, 3 503, 520

³⁶ George P. Smith, II and Thaddeus J. Burns, 'Genetic Determinism or Genetic Discrimination' (1994- 1995) 11 Journal of Contemporary Health Law and Policy 23. See also Jon Beckwith, Foreword: The Human Genome Initiative: Genetics' Lighting Rod (1991) 17 American Journal of Law and Medicine 1, 2. See Maxine Singer and Paul Berg, 'Genes and Genome: A Changing Perspective (1991).

³⁷ Ricki Lewis, *Human Genetics: The Basics* (Routledge 2011)

³⁸ Thomas D. Gelehrter, Francis S. Collins and David Ginsburg, *Principles of Medical Genetics* (2nd edn, Williams & Wilkins 1998) 9

³⁹ Eric Mills Holmes, 'Solving the Insurance/ Genetic Fair/ Unfair Discrimination Dilemma in Light of the Human Genome Project' (1996- 1997) 85 Kentucky Law Journal 3 503, 521- 522

⁴⁰ Ibid at 520

height, eye colour and hair colour, one may also inherit diseases and disorders, such as heart disease, diabetes, and different types of cancer. Certain hereditary diseases can be explained by a single gene, or monogenic defect. However, the majority of hereditary diseases are multifactorial and are caused by the interaction of environmental factors and numerous abnormal genes. The following section will give an overview of the different types of genetic disorders. It is generally acknowledged that there are four classes of genetic disorders: chromosomal disorders, single- gene disorders, multifactorial disorders and mitochondrial disorders.⁴¹

Chromosomal genetic disorders can occur when an individual possesses the incorrect number of chromosomes.⁴² Chromosomal disorders therefore arise if the number or structure of any of an individual's forty- six chromosomes is abnormal, which leads to physical or mental maldevelopment. A typical example of a chromosomal genetic disorder is Down Syndrome, and individuals born with Down Syndrome have three copies, as opposed to two copies of chromosome "twenty- one."⁴³ Chromosomal abnormalities can also arise during the division of cells, with the result that additional or reduced chromosomal material occurs in the cells. In addition, the chromosomal material might be reorganised in these new cells with the result that the balance may be upset, and maldevelopment may occur.⁴⁴

Mitochondrial genetic disorders are genetic disorders caused by mutations in non- chromosomal DNA located within the mitochondria.⁴⁵ Mitochondria "are a subcompartment of the cell bound by a double membrane"⁴⁶ and are found specifically in each cell's cytoplasm. They constitute structures within cells that convert the energy from food into a form that cells can use. Although most DNA is packaged in chromosomes within the nucleus, mitochondria also have a small amount of their own DNA and this genetic substance is known as mitochondrial DNA or mtDNA. Mitochondria DNA is therefore different from regular DNA in

⁴¹ Shannyn C. Riba 'The Use of Genetic Information in Health Insurance: Who will be Helped, who will be harmed and the possible long- term effects' (2007) 16 Review of Law and Social Justice 2 470, 473. See also Human Genome Project Information: http://ornl.gov/sci/techresources/Human_Genome/medicine/assist.shtml (accessed 7 January 2013)

⁴² Thomas D. Gelehrter, Francis S. Collins and David Ginsburg, *Principles of Medical Genetics* (2nd edn, Williams & Wilkins 1998) 3

⁴³ Individuals with Down syndrome have three rather than two copies of chromosome "twenty- one." For further discussion see Stylianos E. Antonarakis, 'Chromosome 21 and Down Syndrome: From Genomics to Pathophysiology' (2004) 5 Nature Reviews: Genetics 725

⁴⁴ Eric Mills Holmes, 'Solving the Insurance/ Genetic Fair/ Unfair Discrimination Dilemma in Light of the Human Genome Project' (1996- 1997) 85 Kentucky Law Journal 3, 503, 528

⁴⁵ Marni J Falk and Neal Sondheimer, 'Mitochondrial genetic disease' (2010) 22 Current Opinion in Pediatrics 6 711

⁴⁶ Patrick F Chinnery, Eric A Schon, 'Mitochondria: Neuroscience for Neurologists' (2003) 74 Journal of Neurology and Neurosurgery Psychiatry 1188, 1189

terms of its structure and function.⁴⁷ The occurrence of mitochondria in the ovum facilitates the inheritance of disorders, through the mother, to future generations.⁴⁸ By way of maternal inheritance, a mother with a mitochondrial genetic mutation therefore passes this affected gene to her children, who will be affected, with varying degrees of severity. Mitochondrial disorders vary in terms of nature and severity, and may express at any age. Some disorders affect just a single organ (for example, the eye in Leber hereditary optic neuropathy⁴⁹), but many affect several organs.⁵⁰

Single gene disorders result from a single gene mutation, which causes the gene to be damaged or missing. It is effectively caused by one abnormally or ineffectively functioning gene. This type of genetic disorder is characterised by a pattern of inheritance.⁵¹ A dominant single gene disorder arises if an abnormality is evident when only one of the chromosomal pair contains the variant gene.⁵² Generally, an individual affected by the dominant single gene disorder had a parent who also had the gene, who also had a parent with the gene, back to the original gene mutation.⁵³ The recessive disorder occurs when an affected individual inherits an abnormal gene from each parent, even in circumstances where neither parent has the particular disorder.⁵⁴ Single gene disorders include muscular dystrophy, cystic fibrosis, sickle- cell anaemia, and Huntington's disease.

Multifactorial (or polygenic) genetic disorders are the largest group of genetic disorders. They are caused from an interaction of complex factors including, multiple genetic mutations, as well as environmental factors, such as diet and lifestyle.⁵⁵ Common multifactorial genetic disorders include heart disease, diabetes and different types of cancer. Testing for multifactorial genetic disorders will indicate only that a person has a higher than average probability of developing a disease, but it is not certain if and when the condition will manifest.

⁴⁷ For further discussion see, Robert W. Taylor and Doug M. Turnbull, 'Mitochondrial DNA Mutations in Human Disease' (2005) 6 *Nature Reviews: Genetics* 389

⁴⁸ Heidi Chial and Joanna Craig, 'mtDNA and Mitochondrial' (2008) 1 *Nature Education* 2

⁴⁹ PYW Man et al, 'Leber hereditary optic neuropathy' (2002) 39 *Journal of Medical Genetics* (2002) 162. See also Douglas C. Wallace, 'Mitochondrial DNA Mutation Associated with Leber's Hereditary Optic Neuropathy' (1988) 242 *Science*, New Edition 4884, 1427

⁵⁰ Andreea Nissenkorn *et al*, 'Multiple Presentation of Mitochondrial Disorders' (1999) 81 *Archives of Disease in Childhood* 3, 209- 215

⁵¹ Thomas D. Gelehrter, Francis S. Collins and David Ginsburg, *Principles of Medical Genetics* (2nd edn, Williams & Wilkins 1998) 4

⁵² Heidi Chial, 'Mendelian genetics: Patterns of inheritance and single gene disorders' (2008) 1 *Nature Education* 1

⁵³ Eric Mills Holmes, 'Solving the Insurance/ Genetic Fair/ Unfair Discrimination Dilemma in Light of the Human Genome Project' (1996- 1997) 85 *Kentucky Law Journal* 3, 503, 528

⁵⁴ *Ibid*

⁵⁵ Thomas D. Gelehrter, Francis S. Collins and David Ginsburg, *Principles of Medical Genetics* (2nd edn, Williams & Wilkins 1998) 4- 5

Discussion of multifactorial genetic conditions therefore provokes consideration of the interaction between genes and environment on disease expression.⁵⁶

2.2.1 Interaction between genes and environment

It was once hypothesised that “‘You,’ your joys and your sorrows, your memories and your ambitions, your sense of personal identity and free will, are in fact no more than the behavior of a vast assembly of nerve cells and their associated molecules.”⁵⁷ Crick was arguably over zealous in his theory of the influence of genetics in shaping all aspects of human life. Research has shown that an individual’s identity and indeed an individual’s predisposition to disease is a complex interaction between both genes and external environmental factors. Although disease and disability are dictated by genes, they also arise as a result of external factors and lifestyle choices, and the interaction of these factors.⁵⁸ Therefore, it is necessary to highlight the impact of environmental factors upon disease expression.

There are few human diseases that are caused by a single gene operating in isolation. Multiple gene disorders, where several mutated genes are involved in triggering the disorder, are more common (albeit less understood). Examples of multiple gene disorders include breast cancer, asthma, diabetes, heart disease. Common disorders such as heart disease and most cancers arise from an intricate interaction among multiple genes and between genes and factors in the environment. An individual’s lifestyle choices and access to health care are some of the environmental factors that can impact upon the expression of a genetic based disorder. It is important to be aware of the interaction between genes and environment, particularly in considering the predictive value of genetic tests in certain circumstances.⁵⁹

2.2.2 Behavioural traits

Advances in genetic science are also revealing the potential to detect certain behavioural traits and predispositions to certain lifestyle activities. Behaviour manifests as a result of complex traits involving multiple genes that are influenced by a number of other factors.⁶⁰ Researchers have discovered a genetic basis for various types of behaviour. Recent research indicates the

⁵⁶ Roy Deodutta and Dorah Tevfile, ‘Environmental Factors, Genes and Development of Human Cancers (Springer 2010)

⁵⁷ Francis Crick, *The Astonishing Hypothesis: the Scientific Search for the Soul* (Scribner 1994) 3

⁵⁸ Colin S. Diver and Jane Maslow Cohen, ‘Genophobia: What is Wrong with Genetic Discrimination?’ (2000- 2001) 149 University of Pennsylvania Law Review 1440, 1451

⁵⁹ Roy Deodutta and Dorah Tevfile, *Environmental Factors, Genes and Development of Human Cancers* (Springer 2010)

⁶⁰ Cynthia T. Garcia Coll *et al*, *Nature and Nurture: The Complex Interplay of Genetic and Environmental Influences on Human Behaviour and Development* (Lawrence Erlbaum 2004)

existence of the risk- taking gene,⁶¹ and predisposition towards determination and success.⁶² Similarly, in recent times, scientists have detected the genetic basis indicating leadership. The leadership gene, known as rs4950, is associated with individuals taking charge.⁶³ Leadership qualities are also learned and factors such as individual's environment can impact upon the expression of leadership skills. Researchers discovered the gene following analysis of DNA samples from around 4,000 individuals and linking them with details of employment and relationships.⁶⁴ The discovery of this gene may reveal interesting insights into those who are likely to become managers and those likely to remain in a more subordinate role, thereby providing potentially valuable information for employment purposes.

The field of behavioural genetics has the potential to reveal both genetic and environmental influences on normal and abnormal behaviour. In addition, this research can yield benefits in various fields of medicine, particularly psychiatric medicine, in finding the most effective treatment options available, based on an individual's unique genetic make up.

2.3 Genetic Testing

The above analysis provided a basis to discuss genetic testing and what such tests can reveal. The success of the HGP (discussed below) has accelerated the pace of genetic discoveries and resulted in an increase in the practice of genetic testing (for medical and non medical purposes). Genetic tests can be broadly categorised into predictive and diagnostic tests. For the purposes of this debate, the majority of the ethical and legal issues arise out of predictive genetic tests and what they can reveal about an individual's predisposition to disease, disability and behavioural tendencies.

⁶¹ Richard Alleyne 'Why risk taking runs in the family – scientists find gene that makes you gamble', *The Telegraph* (London 6 May 2009). Available at: <http://www.telegraph.co.uk/news/newstopics/howaboutthat/5279222/Why-risk-taking-runs-in-the-family-scientists-find-gene-that-makes-you-gamble.html> (accessed 7 June 2012). See also Scott Shane, 'Explaining Innate Risk- Takers', *Bloomberg Businessweek*, (New York 12 February 2010). Available at: http://www.businessweek.com/smallbiz/content/feb2010/sb20100211_567240.htm (accessed 7 June 2012), citing Scott Shane, *Born Entrepreneurs, Born Leaders – How your Genes Affect your Work Life*, (Oxford University Press 2010). See also Mary Jeanne Kreek *et al* 'Genetic Influences on impulsivity, risk- taking, stress, responsibility and vulnerability to drug abuse and addiction' (2005) 8 *Nature Neuroscience* 11 1450

⁶² Christine Hsu, 'New Findings Suggest that Winners are Born, not Bred', *Medical Daily* (16 May 2012). Available at: <http://www.medicaldaily.com/news/20120516/9908/success-genes-dna-characteristics-failure-twins.htm> (accessed 7 June 2012)

⁶³ Jan Emmanuel De Neve *et al*, 'Born to lead? A twin design and genetic association study of leadership role occupancy' (2013) 24 *Leadership Quarterly* 1 45

⁶⁴ *Ibid*

Technologies that “*identify genetic variations*” are known as genetic tests.⁶⁵ Genetic testing has been defined as “*the analysis of a specific gene, its product or function, or other DNA and chromosome analysis, to detect or exclude an alteration likely to be associated with a genetic disorder.*”⁶⁶ In other words, genetic testing (or screening) is the process of scanning an individual’s genetic make- up to ascertain if the individual has a genetic predisposition to developing or passing on a genetic defect or disease.⁶⁷ Genetic monitoring focuses on the workplace itself or other environments as potentially provoking the expression of certain genetic conditions. It follows that certain genetic conditions are exacerbated by exposure to carcinogens or other toxic chemicals. Genetic monitoring is utilised to improve the working environment and therefore protect employees.⁶⁸ Such testing can be carried out to monitor workplace exposure to hazardous substances, which could predict increased predisposition to future illness or disability.⁶⁹

The technique of genetic testing “*involves examining a person’s DNA for some anomaly that flags a disease or disorder.*”⁷⁰ Genetic testing involves cells from the individual being tested, and generally, the cells used come from blood, saliva, the inside of the cheek, or any other human tissue. Suspected mutations and predisposition to disease can be confirmed by genetic testing before symptoms appear.⁷¹ The objective is that suspected genetic abnormalities and predisposition to disease can be confirmed or denied by genetic testing, before the expression of symptoms. There are a number of different types of genetic tests, used in a variety of scenarios.

Firstly, there are genetic tests that can be used to identify carriers of certain diseases or disorders. Carrier screening to detect Mendelian diseases, involves identifying asymptomatic individuals who carry one copy of a gene for a disease that requires two copies for the disease to be expressed.⁷² Individuals who carry genes do not themselves have a particular disease or disorder and may never develop the disease or disorder, but they nevertheless possess recessive genes and pass them along to future generations. Individuals may therefore decide to

⁶⁵ Eric Mills Holmes, ‘Solving the Insurance/ Genetic Fair/ Unfair Discrimination Dilemma in Light of the Human Genome Project’ (1996- 1997) 85 Kentucky Law Journal 3 505, 523- 524

⁶⁶ Peter S. Harper, ‘What do we mean by genetic testing?’ (1997) 34 Journal of Medical Genetics 749, 749

⁶⁷ Melinda B. Kaufman, ‘Genetic Discrimination in the Workplace: An Overview of Existing Protections’ (1999) 30 Loyola University Chicago Law Journal 393, 397- 398

⁶⁸ Kirke D. Weaver, ‘Genetic Screening and the Right not to Know’ (1997) 13 Issues in Law and Medicine 243, 246

⁶⁹ Melinda B. Kaufman, ‘Genetic Discrimination in the Workplace: An Overview of Existing Protections’ (1999) 30 Loyola University Chicago Law Journal 393, 398- 399

⁷⁰ Paul Steven Miller, Is there a pink slip in your genes? Genetic discrimination in the workplace’ (2000) 3 Journal of Health Care Law and Policy 2 225, 229. See also Denise K. Casey, ‘What can the new gene tests tell us?’ (1997) 36 The Judges Journal 14, 16

⁷¹ Peter S. Harper, ‘What do we mean by genetic testing?’ (1997) 34 Journal of Medical Genetics 749

⁷² Neil A. Holtzman *et al*, ‘Predictive Genetic Testing: From Basic Research to Clinical Practice’ (1997) 278 Science 602, 603

undergo such genetic testing where there is a family history of a particular disease, if they have symptoms of a genetic disorder or if they are concerned about passing on genetic disorders to their children. These genetic tests are therefore particularly relevant in the area of reproduction.⁷³

Preimplantation genetic diagnosis is another type of genetic technology, which is particularly relevant in reproduction and which involves screening embryos. It is a technique used to identify genetic abnormalities in embryos created through in vitro fertilisation.⁷⁴ It is noted that this practice potentially produces many ethical concerns in deciding in what circumstances to continue with pregnancy and arguably provokes ethical issues such as the value and dignity of all individuals.

Additional genetic tests are used in the context of reproduction. Prenatal diagnostic testing involves testing the foetus before birth to determine whether the foetus has certain abnormalities, including inherited or random genetic mutations. Genetic disorders such as cystic fibrosis and sickle cell anaemia can be tested at this stage. Newborn screening is a technique used on new born babies to test for a range of genetic disorders, such as the “*heel prick test*.”⁷⁵ These technologies generally identify treatable genetic disorders in babies, usually prior to the manifestation of conditions, many of which are metabolic disorders.⁷⁶ These tests are advantageous from the perspective of earlier diagnosis, with the potential of earlier intervention and prevention.

There are also many predictive genetic tests available for adult-onset disorders. These tests are particularly relevant for the purposes of this thesis. Presymptomatic testing is a type of test for predicting adult-onset disorders, such as Huntington’s disease, and other monogenic genetic conditions.⁷⁷ There is also presymptomatic testing for predicting the risk of developing adult-onset disorders such as Alzheimer’s disease or different types of cancer. These are multifactorial genetic conditions and such genetic testing detects the presence of genes indicating a probability of an individual expressing the condition. As well as predictive genetic testing technologies, genetic testing can also be used as an effective diagnostic mechanism. Confirmational diagnosis of a symptomatic individual may be carried out following the suspected presence of a genetic condition or disease.⁷⁸ In these circumstances, a family history of a genetic condition may be confirmed by undergoing a genetic test.

⁷³ Callum J. Bell *et al*, ‘Carrier Testing for Severe Childhood Recessive Diseases by Next- Generation Sequencing’ (2011) 3 *Science Translational Medicine* 65 1

⁷⁴ Karen Sermon *et al*, ‘Preimplantation genetic diagnosis’ (2004) 363 *The Lancet* 1633. See also Peter Braude *et al*, ‘Preimplantation genetic diagnosis’ (2002) 3 *Nature Reviews* 941

⁷⁵ Stefan Timmermans and Mara Buchbinder, *Saving Babies – The Consequences of Newborn Genetic Screening* (University of Chicago Press 2013) 7

⁷⁶ *Ibid*

⁷⁷ Peter S. Harper, ‘What do we mean by genetic testing?’ (1997) 34 *Journal of Medical Genetics* 749, 750

⁷⁸ *Ibid*

Forensic or identity testing is another type of genetic test, that is generally utilised as an investigative tool from a legal and criminal justice perspective. Such testing is also used in the context of paternity and maternity suits, as confirmation of genetic lineage.⁷⁹

In terms of accessibility, the cost of genetic testing can range from hundreds to thousands of dollars or euros, depending on the nature of the genes and the numbers of mutations tested. Currently, more than 1,000 genetic tests are available, which can test for a range of genetic based illnesses and predisposition to potential illnesses.⁸⁰ In the clinical context, it is also noted that informed consent is required before an individual undergoes genetic testing. It is also generally recommended that an individual undergo genetic counselling, which will help the individual, (and the family) understand genetic disorders, the implications of genetic testing and the various options available.⁸¹ Most genetic tests provide only an estimated risk for developing the disorder and therefore it is acknowledged that the accuracy of genetic testing is limited.

2.3.1 Accuracy of genetic testing

This thesis acknowledges the limitations of genetic science. It is important at this juncture to refer to the accuracy of genetic testing and the general perception of such testing. There is a common belief is that genetic technologies “*are always accurate, highly predictive and capable of identifying an individual’s or offspring’s inevitable pre- destination of future disability. The facts are diametrically opposed to this common belief.*”⁸² Although science is advancing rapidly, the majority of genetic tests are still uncertain and lack accuracy. Predicting the nature and severity of disease based upon an identified gene is challenging.⁸³ The probability that a genetic flaw will manifest in a particular disease or disorder in the future is dependent on a variety of complex factors. Therefore the scientific accuracy and validity of genetic tests are limited.

In this context, it is important to refer to the distinction between multifactorial genetic conditions and monogenic conditions. In the case of the majority of genetic conditions (for example, multifactorial genetic conditions), the time of onset, severity of the condition and impact and management of treatment varies greatly.⁸⁴ For example, individuals who are diagnosed with the gene for

⁷⁹ Ibid

⁸⁰ See <http://ghr.nih.gov/handbook/testing?show=all> (accessed 25 July 2013)

⁸¹ Peter S. Harper, ‘What do we mean by genetic testing?’ (1997) 34 Journal of Medical Genetics 749, 751

⁸² Lawrence O. Gostin, ‘Genetic Discrimination: The Use of Genetically Based Diagnostic and Prognostic Tests by Employers and Insurers’ (1991) 17 American Journal of Law and Medicine 1 & 2 109, 113

⁸³ Neil A. Holtzman *et al*, ‘Predictive Genetic Testing: From Basic Research to Clinical Practice’ (1997) 278 Science 602, 604

⁸⁴ Ibid at 114

Huntington's disease⁸⁵ will develop, with a "*chilling certainty*", the fatal disorder generally around middle age.⁸⁶ On the other hand, the detection of the breast cancer gene, (BRCA1 mutation) has a more uncertain predictive value. An individual who is diagnosed with the breast cancer gene has a reasonably high probability of developing breast cancer. This probability is also influenced by possible history of breast cancer in the family. Similarly, a genetic test result indicating a predisposition to heart disease, is dependent upon a number of external factors such as lifestyle, diet and other potential genetic markers.⁸⁷

Therefore, the predictive value of genetic tests is somewhat limited because generally they reveal merely the possibility that the person may develop the trait, disease or disorder in the future and are not certain indicators that symptoms will develop. Therefore, possessing a genetic mutation does not necessarily lead to the expression of the disease or disorder.⁸⁸ In the future, it is anticipated that genetic tests will become more sophisticated, and will be able to predict the expression and onset of disease more accurately, as well as the degree to which environmental factors contribute towards disease. It is necessary to be aware of not only the potential benefits of genetic technologies, but also the limitations of such technologies at this stage.

2.3.2 Genetic information

For the purpose of this thesis, it is necessary to clarify the concept of genetic information.⁸⁹ It is important to point out that an individual's genetic information can also be revealed without the use of genetic tests. For example, one may discover such information through insights obtained from their medical history, as well as details of family medical history.⁹⁰ Family medical history has traditionally been a reliable source of genetic information and individuals can obtain considerable knowledge from analysis of the medical history of one's blood-relatives. Therefore, when discussing the concept of genetic information and use of such information, it is necessary to refer to both the results of genetic tests as well as information gained from family medical history. As will be highlighted throughout this thesis, genetic information, gained from test results, as well as from family history, can potentially be misused.

⁸⁵ For further details see www.huntingtons.ie (accessed 25 July 2013)

⁸⁶ Paul Steven Miller, 'Is there a Pink Slip in my Genes? Genetic Discrimination in the Workplace' (2000) 3 Journal of Health Care Law and Policy 2 225, 231

⁸⁷ Francois Cambien and Laurence Tiret, 'Genetics of Cardiovascular Diseases – From Single Mutations to the Whole Genome' (2007) 116 Circulation 1717 (<http://circ.ahajournals.org>) (accessed 25 July 2013)

⁸⁸ Paul Steven Miller, 'Is there a Pink Slip in my Genes? Genetic Discrimination in the Workplace' (2000) 3 Journal of Health Care Law and Policy 2 225, 229. See also National Cancer Institute United States Department of Health and Human Services, 'Understanding Gene Testing (1995) 4, 5

⁸⁹ The terms "genetic information" and "genetic data" are used interchangeably in this thesis.

⁹⁰ Ibid at 231

2.3.3 Benefits of genetic testing

These technologies may potentially enhance health care and medical treatment.⁹¹ By taking a genetic test, an individual can discover what genes he/she may have that may potentially indicate predisposition to a range of conditions and diseases. Genetic technology may offer the prospect of being able to detect the onset of future disabilities, thereby highlighting genetic predispositions to disability. In addition to identifying genetic markers for disease and disability, genetic testing can also identify potential behavioural and personality traits in individuals, with potentially vast benefits in the field of psychiatric medicine, as noted above.

Genetic testing offers opportunities for individuals to be fully informed as to their genetic status, so that they can effectively manage their health care and treatment plans. This facilitates the practice of personalised medicine, a concept which effectively tailors health care to an individual, based on his/ her unique genetic make up and individual constitution.⁹² These technologies also improve the health care profession generally, by providing health care professionals with detailed information about a patient's genetic make up, thereby enabling more effective diagnosis and treatment. Genetic technologies can offer the treatment and possible cures for diseases at an earlier stage.⁹³ Genetic testing and its potential arguably enhances patient autonomy and self-determination, increasing self-awareness and ultimately enabling patients to make more informed decisions in relation to their health care, and the health care of their family members. From a societal perspective, genetic testing offers significant public health benefits.

Genetic testing and other genetic technologies advanced throughout the 20th century. The modern genetic revolution began in the 21st century and was accelerated exponentially with the success of the HGP. The next section will examine the HGP in further detail.

3. The Human Genome Project

⁹¹ See generally Saini Rajiv et al., 'Genetic Screening: The vista of genomic medicine' (2011) 3 Journal of Pharmacy and Bioallied Sciences 1 109. For further discussion of the practice of genetic testing in the clinical setting, see Steven W.J. Lamberts and Andre G. Uitterlinden, 'Genetic Testing in Clinical Practice' (2009) 60 Annual Review of Medicine 431- 442

⁹² Leslie P. Francis, 'You are Born with Your Genes: Justice and Protection against Discrimination in the Use of Genetic Information' (2010) 77 Mount Sinai Journal of Medicine 188. See also Margaret A. Hamburg and Francis S. Collins, 'The Path to Personalized Medicine' (2010) 363 New England Journal of Medicine 4 301

⁹³ David Batty, 'Breast cancer treatment gets boost', *The Guardian* (London 19 April 2012) <http://www.guardian.co.uk/science/2012/apr/19/breast-cancer-treatment-boost> (accessed 19 April 2012)

Although the 20th century witnessed extraordinary discoveries in the field of genetics, there was still a great deal that was unknown about the genetic basis of disease. In exploring the history of genetic science, it is necessary to highlight the importance of the HGP as a key milestone in the genetic science revolution. This section will provide an overview of the HGP, highlighting its relevance in this area. The HGP was a 13- year quest to sequence the human genome, with a view to gaining a greater understanding of disease and genes. It has been the first significant endeavour of the biological and medical research communities into what has been described as “*big science*.”⁹⁴ The HGP was one of the largest and most significant scientific projects embarked upon in both the US and internationally. The following sections will look more closely at the reasons behind the HGP, its goals and its impact on the area of genetics.

3.1 Motivation behind the Human Genome Project

In response to the need to develop genetic technologies and to help tackle genetic disease, there was a recognised need for a concerted international effort to unravel the human genome and to enable geneticists to continue to chart the genetic basis of a wide range of diseases. In 1988 the US Congress funded the National Institutes of Health (NIH) and the Department of Energy (DOE) for a project to explore the human genome.⁹⁵ In 1988 James Watson took the lead in a new NIH component of the effort, which became a joint NIH- DOE project.⁹⁶ The HGP officially began in 1990.

Additionally, many other countries and the EU established human genome research programmes, including Australia, Brazil, Canada, China, Germany, Israel, Italy, Japan, Korea, Mexico, Russia and the United Kingdom.⁹⁷ According to the NIH, the general objective of this multi- disciplinary global initiative was to discover fundamental information needed to further scientific understanding of human genetics and of the role of genes in health and disease.⁹⁸ This \$3 billion project was expected to take 15 years to complete, but rapid technological advances, together with competition from private endeavours accelerated the process and the completion date was brought forward by two years.⁹⁹

⁹⁴ Francis S. Collins, Michael Morgan, Aristides Patrinos, ‘The Human Genome Project: Lessons from Large Scale Biology’ (2003) 300 Science, 286, 286

⁹⁵ The Human Genome Project is part of the National Human Genome Research Institute, at the National Institutes of Health (NIH) – see website: <http://www.genome.gov/10001772> (accessed 12 April 2012)

⁹⁶ Francis S. Collins, Michael Morgan, Aristides Patrinos, ‘The Human Genome Project: Lessons from Large Scale Biology’ (2003) 300 Science, 286, 286

⁹⁷ Ibid

⁹⁸ Oak Ridge Laboratories, Human Genome Project Information, Project 5 Year Research Goals 1998-2003, at http://www.ornl.gov/TechResources/Human_Genome/hg5yp/ (accessed 25 July 2013)

⁹⁹ Heidi Chial, ‘DNA Sequencing Technologies Key to the Human Genome Project’ (2008) 1 Nature Education 1

3.2 Goals of the Human Genome Project

The HGP had a number of very specific goals (the majority of which were achieved ahead of schedule and under budget). In April 1990 the NIH and DOE published a five- year plan.¹⁰⁰ The principal goal was to map and sequence the twenty- four chromosomes that contain the complete genetic contents of a human cell – in other words, the aim was to sequence the entire human genome.¹⁰¹ The basic idea of the map is that it establishes specific markers throughout the genome that can be used as reference points to pinpoint specific genes on chromosomes. The project effectively creates a map of the entire genetic structure of the human species. The map is then used to develop new ways to treat, cure and prevent diseases and disorders. According to Collins and Monsura, the objective of the HGP was “*to unravel some of the mysteries of disease by unravelling the thread of DNA present in nearly every cell in our bodies.*”¹⁰²

One of the most important goals was to identify and address the potential ethical, legal and social issues.¹⁰³ The HGP therefore recognised its responsibility to consider how the data generated will impact society.¹⁰⁴ Indeed one of the main goals of the HGP’s was to “*develop programs addressed at understanding the ethical, legal and social implications of the Human Genome Project, identify and define the major issues, and develop initial policy options to address them.*”¹⁰⁵ This segment of the HGP had a number of purposes, including, anticipating the consequences for individuals and society in general of undertaking this project, addressing the ethical and legal dilemmas of sequencing the human genome, engaging the public in this debate and formulating policy options to ensure the appropriate use of these new technologies.¹⁰⁶ The inclusion of a budget to address these issues is a recognition that although scientific advances have enormous potential benefit, there is also a potential for misuse.

¹⁰⁰ Understanding Our Genetic Inheritance: The US HGP, The First Five Years: Fiscal Years 1991-1995, available at http://www.nhgri.nih.gov/HGP/HGP_goals/5yrplan.html (accessed 3 May 2013)

¹⁰¹ For further details, see National Human Genome Research Institute, ‘From Maps to Medicine: About the Human Genome Research Project’, available at http://www.nhgri.nih.gov/Policy_and_public_affairs/Communications/Maps_to_medicine/about.html (accessed 7 January 2013)

¹⁰² Francis S. Collins, Monique K. Monsura, The Human Genome Project: Revealing the Shared Inheritance of All Humankind (7th Biennial Symposium on Minorities, the Medically Undeserved and Cancer) at 222

¹⁰³ Francis S. Collins, Michael Morgan, Aristides Patrinos, ‘The Human Genome Project: Lessons from Large Scale Biology’ (2003) 300 Science 286, 289

¹⁰⁴ The group that worked on these issues is referred to as the Ethical, Legal, Social Implications (ELSI) program. See <http://www.nhgri.nih.gov/ELSI>

¹⁰⁵ Understanding Our Genetic Inheritance: The US HGP, The First Five Years: Fiscal Years 1991-1995, available at http://www.nhgri.nih.gov/HGP/HGP_goals/5yrplan.html (accessed 3 January 2012)

¹⁰⁶ Ibid.

Collectively, these goals would act to contribute towards a greater understanding of the genetic basis of disease, for the medical and scientific community and for society as a whole, as well as recognising the need to acknowledge the corresponding ethical and legal dilemmas that arise. In 1993, rapid advancements in the HGP led the NIH and the DOE to amend their initial goals.¹⁰⁷ Following unprecedented success at the end of the revised five- year plan, a new plan was introduced in 1998 that further refined the HGP's goals.¹⁰⁸

3.2.1 Private versus public human genome sequencing

As highlighted, the HGP was completed ahead of schedule and under budget. A major contributing factor behind this success was the private endeavour to sequence the human genome. The private effort was spearheaded by Celera Genomics, which began competing against the public HGP in 1998¹⁰⁹ (co-founded by Applera Corporation (then called Perkin Elmer Corporation) and Dr. J. Craig Venter). Celera's mission was to generate and commercialise genomic information.¹¹⁰ Specifically, it aimed to sequence the human genome and provide its future clients with early access to the resulting data. The firm aimed to complete the sequencing of the human genome quicker (within three years) and at a reduced cost of the public project (\$300 million versus \$3 billion).¹¹¹ Celera had attempted to seek intellectual property rights in the genes, however in March 2000, it was announced that the genome sequence could not be patented, and should be made freely available to all researchers. This announcement had a detrimental impact on Celera's stock and also had an overall negative effect on the Nasdaq, which was dominated by biotechnology.¹¹² The private versus public competition provoked the publicly funded project to alter their strategy in order to accelerate progress. Indeed, the private element to this competition was applauded by those in the public project, as enhancing the completion of the endeavours.¹¹³

3.3 The completion of the Human Genome Project

¹⁰⁷ Francis Collins and David Gallas, A New Five Year Plan for the US Human Genome Program, available at http://www.nhgri.gov:80/HGP/HGP_goals/plan.html (accessed 25 July 2013)

¹⁰⁸ Francis Collins et al, 'New Goals for the U.S. Human Genome Project: 1998- 2003, available at <http://www.sciencemag.org/cgi/content/full/282/5389/682> (accessed 12 December 2012)

¹⁰⁹ For further details: <https://www.celera.com/> (accessed 22 August 2012)

¹¹⁰ Marc A. Shampo and Robert A. Kyle, 'J. Craig Venter – The Human Genome Project' (2011) 86 Mayo Clinic Proceedings 4 26

¹¹¹ Kenneth G. Huang, Fiona E. Murray, 'Entrepreneurial Experiments in Science Policy: Analyzing the Human Genome Project' (2010) Research Policy 39.5, 567- 582. (<http://hdl.handle.net/1721.1/65078>)

¹¹² Tom Reynolds, 'Genome Data Announcement Fuels Stock Plunge, Misunderstanding', (2000) 92 National Journal of Cancer Institute 8 594

¹¹³ Francis Collins, Testimony before the Health, Education, Labor and Pensions Committee, United States Senate, Hearing on Genetic Information in the Workplace (20 July 2000) <http://www.genome.gov/10001380>

In June 2000, it was confirmed that a rough draft of the human genome was completed a year ahead of schedule, which was described as “*the most wondrous map ever produced by mankind*”, developed in this “*greatest age ever known*.”¹¹⁴ In 2003 it was announced that they had successfully mapped the human genome, two years ahead of schedule.¹¹⁵ In April 2003 researchers publicly released the results of the HGP. GenBank holds all of the genetic information deposited by the international contributors to the HGP, in a publically available and accessible forum.¹¹⁶ As a publically available resource, the information provided by GenBank is a valuable tool in promoting genetic discoveries and facilitating knowledge sharing.

3.4 Consequences of the Human Genome Project

The HGP culminated in the successful sequencing and publication of the draft sequence of the human genome in 2001 and the complete sequence in 2003. The HGP has generated great interest in the scientific and medical communities. This is due primarily to the promise of facilitating knowledge that could revolutionise the manner in which disease is diagnosed and treated. Following the sequencing of the human genome and the many genetic puzzles which the HGP solved, scientists have been provided with information that is proving vital to developing an enhanced understanding of genetics. The HGP was applauded as having “*three main advantages, namely improved diagnostics, new approaches to the prevention of disease, and gene enhancement therapy*.”¹¹⁷

As a result of the success of the HGP and subsequent advancing technology, genetics is playing an increasingly important role in the diagnosis, prevention, and treatment of diseases. Genetic advances will help facilitate the preferable approach to health care, from a preventative, diagnostic or therapeutic perspective. Genomics, the study of the genomes of organisms, including the application of genetic science, is quickly emerging as a key area of science in clinical medicine and diagnosis, highlighting the practical application of this research.¹¹⁸ It has also resulted in a greater understanding of human disease and disability, and the influence of genes, environment and other factors. The success of the HGP offers great opportunities to predict an individual's

¹¹⁴ Remarks by the President Bill Clinton, Tony Blair, Dr Francis Collins, and Dr Craig Venter, on the Completion of the First Survey of the Entire Human Genome Project, White House Press Release, 26 June 2000, White House website, available online at

http://clinton3.nara.gov/WH/EOP/OSTP/html/00628_2.html (accessed 22 August 2012)

¹¹⁵ For further details about the Human Genome Project and its success, see

<http://www.genome.gov/10001772> (accessed 12 April 2012) and <http://www.genome.gov/> (accessed 12 April 2012). See also

http://www.ornl.gov/sci/techresources/Human_Genome/project/timeline.shtml

¹¹⁶ For further details, see website: <http://www.ncbi.nlm.nih.gov/genbank/> (accessed 22 August 2012).

¹¹⁷ Deirdre Madden, *Medicine, Ethics and the Law*, (2nd ed, Bloomsbury Professional 2011) 292

¹¹⁸ Eric S. Lander, ‘The New Genomics: Global Views of Biology’ (1996) 274 *Science* 5287 536

propensity to develop certain diseases and disabilities.¹¹⁹ This has led to more sophisticated genetic tests and an increase in the number of tests available.

In illustrating the success of the HGP, it is observed that genetic advances have revolutionised the diagnosis and treatment of different types of cancer, particularly breast cancer. The discovery of the BRCA1 and BRCA2 genes associated with breast cancer and ovarian cancer, have led to earlier diagnosis and further treatment options.¹²⁰ An individual's risk of developing breast and/or ovarian cancer is greatly increased if they have the BRCA1 or BRCA2 mutation. In addition, individuals who carry these mutations may be at increased risk of developing other certain cancers.¹²¹ Genetic tests have been designed to test for BRCA1 and BRCA2 mutations and several options are therefore available for monitoring risk in individuals (and family members) who test positive for the BRCA1 or BRCA2 mutation.¹²² One option is surveillance, with a view to detecting cancer early. Methods of surveillance include mammograms and breast exams, the goal of which is to find the cancer early, when it is most treatable, resulting in earlier detection and intervention. Another, more radical option available to those who test positive with these genes is prophylactic surgery, with a view to reducing the risk of developing cancer.¹²³

In addition, there have been major advances in recent years enhancing the diagnosis of breast cancer and also the potential for reoccurrence of breast cancer. Based on recent research, it is thought that in the future, a simple blood test could be a more effective way to test for the early signs of breast cancer than using mammograms. This research indicates that a blood test can detect the early signs of cancer; meaning individuals could have a regular blood test rather than breast screening.¹²⁴

Further, recent research is indicating that a simple blood test could predict if certain types of breast cancer are likely to re- occur following treatment.¹²⁵ By facilitating an advance warning this would avoid unnecessary treatment with anti-cancer drugs. Scientists claim they discovered a 'genetic marker' that could be

¹¹⁹ Lawrence O. Gostin, 'Genetic Discrimination: The Use of Genetically Based Diagnostic and Prognostic Tests by Employers and Insurers' (1991) 17 American Journal of Law and Medicine 1 & 2 109, 113

¹²⁰ Steven A. Narod and William D. Foulkes, 'BRCA1 and BRCA2: 1994 and Beyond' (2004) 4 Nature Reviews Cancer 665

¹²¹ Ibid

¹²² Ibid

¹²³ Ibid

¹²⁴ Press Release, Cancer Research UK, 'A simple blood test could be used to detect breast cancer' (2 October 2012) <http://www.cancerresearchuk.org/cancer-info/news/archive/pressrelease/2012-10-02-breast-cancer-blood-test> (accessed 15 May 2013)

¹²⁵ Yadak Sapkota et al. 'Germline DNA Copy Number Aberrations Identified as Potential Prognostic Factors for Breast Cancer Recurrence' (2013) Public Library of Science ONE 8(1): e53850.

doi:10.1371/journal.pone.0053850

<http://www.plosone.org/article/info%3Adoi%2F10.1371%2Fjournal.pone.0053850>

the key to predicting which patients' cancer will re- occur at a later stage.¹²⁶ Reflecting the aim of personalised medicine, treatment strategies could be tailor-made for individuals based on their genetic make-up and ascertain their susceptibility to breast cancer recurrence. These genetic advances therefore offer the prospect of longer lives and increased survival rates.

The discovery of these genes, the genetic tests that have been developed, and the treatment options available have contributed towards more effective diagnosis and treatment of breast and ovarian cancer. The result is a higher rate of survival, and an overall improvement in public health. It is anticipated that in the future, certain types of cancer will no longer be described as fatal, life-threatening diseases, but as chronic illnesses that are easily detected, treated or prevented. The success of the HGP, together with the growing interest in the area of genetics has led to an significant increase in the practice of genetic testing, for medical and other reasons. The HGP laid the foundations for a 21st century revolution in genetic research that offers longer, healthier futures for all individuals. The next section will explore these advances in greater detail and illustrate the potential for the future of genetics.

4. The future of genetic technology

The success of the HGP has led to unprecedented advances in the understanding of disease.¹²⁷ Genetic discoveries and technological advances have introduced a new age in genetic exploration. These advances have resulted in an increase in the practice of genetic testing, for diagnostic, predictive and other purposes.¹²⁸ Such advances have facilitated the development of more sophisticated genetic tests, as well as drug treatments and diagnostic techniques. These technologies facilitate the practice of personalised medicine, which is likely to become a routine element of health care in the future. This section will explore recent advances in genetic technologies and illustrate the potential to improve health care.

4.1 Whole Genome Sequencing

Whole genome sequencing and its application is quickly developing as technology advances. Whole genome sequencing is a process that determines the complete DNA sequence of an organism's genome at a single time.¹²⁹ To recap, a genome can be described as the sum of the genetic information that is

¹²⁶ Ibid

¹²⁷ See generally Francis S. Collins, Victor A. Mc Kusick, 'Implications of the Human Genome Project for Medical Science' (2001) 285 *Journal of the American Medical Association* 5 540

¹²⁸ Chad Terhune, 'Spending on genetic tests is forecast to rise sharply by 2021' *Los Angeles Times*, (Los Angeles 12 March 2012). Available at: <http://articles.latimes.com/2012/mar/12/business/la-fi-genetic-test-20120312>

¹²⁹ Pauline C. Ng and Ewen F. Kirkness, 'Whole Genome Sequencing' (2010) 628 *Genetic Variation – Methods in Molecular Biology* 215

contained in cells and passed from one generation to the next. This process involves sequencing all of an organism's chromosomal DNA as well as the DNA contained in the mitochondria.¹³⁰ This practice provides a whole picture of an individual's genome, including susceptibility to disease and disability.

It is anticipated that this technology will offer great benefits that will be used in medicine, by facilitating more effective diagnostics for genetically based disease.¹³¹ It may therefore operate as a diagnostic technique of children with, for example, intellectual disability, autism, and developmental delays of an unknown cause. Using genetics as a diagnostic tool in these circumstances will provide for earlier diagnosis, earlier intervention and consequently better treatment and options for the individual, thereby enhancing quality of life and patient choice.¹³² In addition, this technology can also be used in identifying individuals who might be sensitive to certain drugs.¹³³ This is effectively a form of pharmacogenetics, (discussed below). It is also anticipated that genomes will be sequenced to identify the best and most effective treatment regimes for different types of cancers.¹³⁴ Whole genome sequencing therefore facilitates personalised medicine. In addition, whole genome sequencing may be used for reproductive purposes. Couples may decide to sequence their genomes before having children, to gain insights into potential genetic abnormalities before making an informed reproductive decision.

With the decreasing cost of whole genome sequencing, it is likely that this technology will become more routine in health care in the future.¹³⁵ In January 2012, Life Technologies announced that it had developed technology that could sequence the whole genome for just over \$1,000.¹³⁶ It was also reported in March 2012 that the human genome can now be sequenced for \$1,000, and inevitably this will continue to decrease in price.¹³⁷ It is anticipated that it will one

¹³⁰ Ibid

¹³¹ Jay Shendure *et al*, 'Advanced Sequencing Technologies: Methods and Goals' (2004) 5 *Nature Reviews: Genetics* 335, 342

¹³² Ciaran Morrison, 'The Evolution of Genetic Science and Technology' at 'Genetic Discrimination – Transatlantic Perspectives on the Case for a European Level Legal Response' (6 March 2012, Brussels, Belgium)

¹³³ Ibid

¹³⁴ Gina Kolata, 'In Gene Sequencing Treatment for Leukemia, Glimpses into the Future', *New York Times* (New York 7 July 2012). Available at: <http://www.nytimes.com/2012/07/08/health/in-gene-sequencing-treatment-for-leukemia-glimpses-of-the-future.html?pagewanted=all> (accessed 20 August 2012). See also Elaine R. Mardis and Richard K. Wilson, 'Cancer Genome Sequencing: A Review' (2009) 18 *Human Molecular Genetics*, Review Issue 2 163

¹³⁵ Ibid, Kolata

¹³⁶ Matthew Herper, 'Not Quite the \$1000 Genome, But Maybe Close Enough' *Forbes* (New York 10 January 2012). Available at: <http://www.forbes.com/sites/matthewherper/2012/01/10/not-quite-the-1000-genome-but-maybe-close-enough/>

¹³⁷ John Markoff, 'Cost of Gene Sequencing Falls, Raising Hopes for Medical Advances', *New York Times* (New York 7 March 2012). Available at: <http://www.nytimes.com/2012/03/08/technology/cost-of-gene-sequencing-falls-raising-hopes-for-medical-advances.html?pagewanted=all& r=0> (accessed 22 July 2013)

day be a powerful diagnostic tool and a means for effective health care management.

The benefits of advancing technology are quickly being realised, not only for individuals, but also for the medical industry and companies specialised in genetic testing and human genome sequencing.¹³⁸ It is also important to point out the corresponding challenges that will inevitably arise in the application of whole genome sequencing. This technique results in a huge amount of information being created about an individual's genetic make-up. Accordingly, a great deal of expertise and knowledge is needed to read and interpret the results as well as acknowledging the inevitable challenges of this practice.¹³⁹ Whole genome sequencing may raise practical considerations particularly where the technique reveals more information than an individual wishes to know.

4.2 Pharmacogenetics

Scientific advances are also facilitating the development of pharmacogenetics.¹⁴⁰ Pharmacogenetics is based on the premise that individuals respond differently to drugs and often vary in their response to the same drug.¹⁴¹ Variation in response to medicines can be based on genetic factors, for example, the genetic make-up of a person influences the process and rate medicines pass through an individual body.¹⁴² As another example of personalised medicine, pharmacogenetics involves tailoring drug treatments to an individual's genetic makeup.

Pharmacogenetics may facilitate greater efficiency as regards expenditure in health care,¹⁴³ as well as safer and more effective use of drugs.¹⁴⁴ The technique can offer benefits, not only for an individual and for the medical fields, but also for the pharmaceutical industry, with the potential to develop more accurate drug treatments with more effective results, thereby promoting efficiency. Indeed,

¹³⁸ A number of companies specialise in whole genome sequencing, for example, <http://www.completegenomics.com/> (accessed 12 December 2012) and <http://www.illumina.com/> (accessed 12 December 2012)

¹³⁹ For further discussion, see Kelly E. Ormond *et al*, 'Challenges in the clinical application of whole-genome sequencing' (2010) 375 *The Lancet* 9727

¹⁴⁰ Trudo Lemmens, 'Selective Justice, Genetic Discrimination, and Insurance: Should we Single Out our Genes in Our Laws?' (2000) 45 *McGill Law Journal* 347, 351. See generally, William E. Evans and Mary V. Relling, 'Moving towards individualized medicine with Pharmacogenomics' (2004) 429 *Nature*, 464

¹⁴¹ Henriette Roscam Abbing, 'Pharmacogenetic data: Mapping the subject's rights' 13 *Good Clinical Practice Journal* (2006) 28, 28

¹⁴² *Ibid*

¹⁴³ Trudo Lemmens, 'Selective Justice, Genetic Discrimination, and Insurance: Should we Single Out our Genes in Our Laws?' (2000) 45 *McGill Law Journal* 347, 351

¹⁴⁴ Allen E. Buchanan *et al*, 'Pharmacogenetics: Ethical Issues and Policy Options' (2002) 12 *Kennedy Institute of Ethics Journal* 1, 3

pharmacogenetics is already being used successfully in clinical practice, and this practice has been particularly effective in the treatment of certain cancers.¹⁴⁵

As with other types of genetic technology, it must be pointed out that drug response depends not only on an individual's genetic structure, but upon a number of other factors as well, including environment, current health, and reaction to drug treatment.¹⁴⁶ Therefore, where pharmacogenetic tests are used, sophisticated clinical judgment is necessary to determine the practical implications of the test results.¹⁴⁷ Education is also key, to fully understand the benefits and limitations of this technology. Advances in pharmacogenetic research and the resulting information it produces also raises a variety of ethical and legal dilemmas that need to be addressed in order to facilitate further developments in the technology. These issues will be discussed further in the next chapter.

4.3 Direct to Consumer genetic testing

In recent years, there has also been a surge in the availability of direct – to – consumer (DTC) genetic testing,¹⁴⁸ illustrating the speed at which genetic science is advancing, and the increasing accessibility of genetic testing. DTC genetic tests are genetic tests that are accessible directly to the consumer. Such genetic tests facilitate access to an individual's genetic information, for example, through mail, or over the Internet. Traditionally, genetic tests have been available only through medical professionals. These tests effectively bypass the requirement for a medical professional in the process of discovering one's genetic status.

Relying on the power of the Internet and recent genetic advances, “*an increasing number of companies are starting to offer health- related genetic testing services directly to the public.*”¹⁴⁹ It is observed that the growing market for DTC genetic testing may promote awareness of genetic diseases, allowing consumers to take control of their genetic health and take a more proactive role in their health care. In this regard, it is observed that these tests “*enhance autonomy and free choice,*

¹⁴⁵ R. Nagasubramanian *et al*, 'Pharmacogenetics in cancer treatment' (2003) 54 Annual Review of Medicine 1 437. See also Marc Ansari and Maja Krajcinovic, 'Pharmacogenomics in cancer treatment defining genetic bases for inter- individual differences in responses to chemotherapy' (2007) 19 Current Opinion in Pediatrics 1 15

¹⁴⁶ Allen E. Buchanan *et al*, 'Pharmacogenetics: Ethical Issues and Policy Options' (2002) 12 Kennedy Institute of Ethics Journal, 1, 4

¹⁴⁷ Ibid

¹⁴⁸ The increase in interest and availability of DTC genetic testing is partly the result of the emergence of several companies, such as: 23andMe <https://www.23andme.com/> (accessed 14 April 2012), deCODEME <http://www.decodeme.com/> (accessed 14 April 2012)

¹⁴⁹ Stuart Hogarth *et al*, 'The Current Landscape for Direct- to- Consumer Genetic Testing: Legal, Ethical and Policy Issues' (2008) 9 Annual Review of Human Genomics and Human Genetics 161, 162. See generally John Lynch *et al*, 'Media Coverage of Direct- to- Consumer Genetic Testing' (2011) 20 Journal of Genetic Counselling 5 486

in that any person who wishes to know about his or her health risks can learn about them, in an accessible manner.”¹⁵⁰ This pattern in health care has “shifted the central role physicians have traditionally played in making medical decisions towards a balance that favours the patient’s particular risk and value preferences.”¹⁵¹

However, such genetic tests, have significant risks and limitations. Indeed, DTC genetic tests are a prime example of advancing genetic technology giving rise to a range of ethical and legal issues. Consumers may make important health care decisions on the basis of inaccurate test results. In the absence of medical guidance and genetic counselling, this may lead to psychological and emotional distress for the individual. Consumers may also experience an invasion of privacy and discrimination if DTC genetic testing companies use their genetic information in an unauthorised manner or if test results are available to third parties. Inadequate regulation in the DTC genetic testing industry may further exacerbate the possibility of risk of harm and abuse of genetic information.

These technologies and new discoveries highlight the speed at which science is developing, and the potential benefits for individuals and for society. The speed at which science is advancing also highlights how genetics is increasingly becoming a part of medical practice.¹⁵² DTC genetic testing, and the emergence of other types of genetic technologies increases the accessibility of genetic tests and contributes towards the avalanche of genetic information that is quickly becoming available to individuals, family members and interested third parties. Legal and ethical issues therefore arise when genetic information is used and misused by third parties, and these issues need to be addressed if science is to continue to flourish.

5. Conclusion

A thorough examination of this topic necessitates an informed understanding of genetic science and the reality of advancing technology. Traditionally, the law has merely reacted to existing problems and waited until an issue needed to be addressed and regulated. However, it is submitted that this emerging interdisciplinary area requires interaction between science and law to effectively

¹⁵⁰ For further discussion, see Rogeer Hoedemaekers, ‘Commercial predictive testing: the desirability of one overseeing body’ (2000) 26 *Journal of Medical Ethics* 282, 284. See also Deepthy Kishore, ‘Test at your own Risk: Your Genetic Report Card and the Direct-to- Consumer Duty to Secure Informed Consent’ (2009- 2010) 59 *Emory Law Journal* 1553

¹⁵¹ Marshall B. Kapp, ‘Patient Autonomy in the Age of Consumer Driven Health Care: Informed Consent and Informed Choice’ (2006) 2 *Journal of Health and Biomedical Law* 1, 2. See also Lucy Modra, ‘Prenatal Genetic Testing Kits Sold at Your Local Pharmacy: Promoting Autonomy or Promoting Confusion?’ (2006) 20 *Bioethics* 254, 263

¹⁵² Jon Cohen, ‘The Human Genome, a Decade Later’, *Technology Review* (January/ February 2011) Available at: <http://www.technologyreview.com/featured-story/422140/the-human-genome-a-decade-later/> (accessed 20 August 2012).

understand the nature and consequences of the issues arising and how to adequately anticipate and address them.

The past century has witnessed great progress in genetics from the discovery of the DNA double helix to the successful completion of the HGP. These and subsequent scientific developments have provoked a worldwide interest in genetics and prompted increasing efforts to learn more about the genetic make-up of human beings, as well as the reasons behind disease and disability. Advancing genetic technology offers the possibility of personalised medicine, which, in the future, may potentially transform health care – through earlier diagnosis, more effective prevention and treatment of disease, and avoidance of drug side effects. It also offers the potential of being able to detect the onset and possible severity of future disabilities. The technology is becoming more prevalent and is being used increasingly in mainstream health care practice, as highlighted in the discussion of breast cancer diagnosis and treatment. The speed at which science is advancing becomes clear, as does the speed at which genetic technologies are becoming more sophisticated and cheaper. This results in a greater availability of genetic testing and genetic information.

This chapter also highlighted some of the limitations of genetic technologies, from the perspective of predicting future disease and disability. It found that the majority of conditions are caused by the complex interaction of genes and environmental factors. In addition, it therefore demonstrated that the majority of genetic tests available yield limited accuracy and statistical validity in terms of predictive value. This may lead to misinterpretation and misuse of such technologies.

As illustrated in this chapter, genetic science and technology is therefore advancing at a rapid pace. Although the HGP and subsequent advances in science have provided the impetus for a revolution in genetic science, it has also therefore generated concerns. Indeed, it is submitted that *“technology is advancing far more rapidly than our ability to legislate or otherwise deal with the moral ramifications of our newfound expertise.”*¹⁵³

These advances in genetic science require an innovative approach towards law and regulation, taking into consideration the reality and speed of technological advances and the subsequent ethical and legal issues that arise from potential misuse of genetic information. The following chapters will look at the various ethical and legal dilemmas arising from use of genetic science, by exploring the various competing and conflicting interests in genetic information and public policy issues arising, from the perspective of the individual and family members, third parties and society as a whole.

¹⁵³ Deborah L. McLochlin, ‘Whose Genetic Information is it Anyway? A Legal Analysis of the Effects that Mapping the Human Genome will have on Privacy Rights and Genetic Discrimination’ (2001) 19 John Marshall Journal of Computer and Information Law 609, 615

Chapter 3: Ethical implications, legal issues and public policy concerns of advancing genetic technology

1. Introduction

While genetic developments promise a new era for healthcare, these developments also bring apprehension and give rise to a myriad of ethical and legal issues. It has been observed that in seeking an appropriate legal response to advances in genetics “*we require laws that are informed by ethical debate, that are morally sound, and that reflect as largely as possible our common societal values.*”¹ It is therefore important to consider the ethical contours of this debate and the legal issues arising, in achieving the appropriate reconciliation between the various competing interests. In particular, questions arise in relation to access to and use of this new information.

This chapter is framed primarily around the theory of the right to know. It is therefore illustrative to look at who has an interest in or ‘right to know’ genetic information and on what basis. It is noted that there are four main groups who potentially have an interest in genetic information and genetic make- up, for a variety of reasons.² Firstly, we ourselves have an interest in knowing our genetic profile. Secondly, family members may have an interest in genetic information, as do spouses and partners. Thirdly, third parties including employers and insurance companies may have an interest in genetic information, primarily from a financial perspective. There are strong commercial interests at stake here. Finally, it is observed that there is a societal interest in genetic information, and in the appropriate application of genetic information. Society has an interest in allowing genetic science and technology to prosper, for the benefit of all, and with a view to improving the efficiency of public health.³

In light of these various interests, questions arise in relation to access to genetic information, the right to know and the tension that is created between the competing rights. This chapter will discuss these interests, with a view to illustrating the tension or conflict between the competing rights. This chapter will also examine the broader public policy concerns which shape this debate, such as the theory of genetic determinism, as well as tendencies toward eugenic policies and the potential creation of a genetic underclass.

¹ Graeme Laurie, ‘Challenging Medical – Legal Norms: The role of autonomy, confidentiality, and privacy in protecting individual and familial group rights in genetic information’ (2001) 22 The Journal of Legal Medicine 1, 2.

² See generally Tuija Takala and Heta Aleksandra Gylling, ‘Who should know our genetic makeup and why?’ (2000) 26 Journal of Medical Ethics 171

³ Ibid at 171

2. An individual's rights – respecting autonomy

An individual has a direct interest in genetic information. Arguably, genetic information is “*the most personal information of all.*”⁴ Genetic information has a unique relationship with the individual, including the fact that it reveals information about an individual's future health status, such information may influence an individual's self-perception, and may potentially stigmatise the person. This direct interest in genetic information incorporates a right to know and a right not to know. In general terms, the right to know can be equated with the right to communicate. It is key to communication and human interaction and this is particularly relevant as regards personal information.⁵ An individual's right to know is an expression of one's inherent right to autonomy. Individuals should be respected as “*autonomous beings who hold views, make choices and take actions based on their personal values and beliefs,*”⁶ and therefore have the right to control genetic information.

Autonomy means “*self-rule, in other words making one's own deliberate decisions.*”⁷ Accordingly, two core elements of autonomy can be identified, firstly, choice, meaning “*to have one's choices respected,*” and secondly, “*non-interference*” from others in making one's choices.⁸ Medical professionals are obliged to respect the choices of their patients even if the choices are potentially harmful. Maintaining autonomy is important in terms of communicating with patients as to their medical position and care, and facilitating their choices.⁹ Allied to the concept of autonomy is the notion of self-determination, which is an individual's right to decide what shall be done to their person.¹⁰ Individuals have the right to decide what is done to their bodies and therefore can make rational decisions, as well as irrational decisions that may have an adverse effect on their health.¹¹

⁴ Graeme Laurie, 'The Most Personal Information of All: An Appraisal of Genetic Privacy in the Shadow of the Human Genome Project' (1996) 10 International Journal of Law, Policy and the Family 74

⁵ Thomas I. Emerson, 'Legal Foundations of the Right to Know' (1976) Yale Law School Faculty Scholarship Series. Paper 2800. Available at: http://digitalcommons.law.yale.edu/fss_papers/2800 (accessed 20 July 2013)

⁶ Nancy E. Kass, 'The Ethical, Legal and Social Issues Concerning the Use of Genetic Tests by Insurers: Toward the Development of Appropriate Public Policy (January 1992) (Available at National Institutes of Health, National Center for Human Genome Research)

⁷ Deirdre Madden, *Medicine, Ethics, and the Law* (2nd edn, Bloomsbury Professional 2011) 45

⁸ Graeme Laurie, *Genetic Privacy: A Challenge to Medico-Legal Norms* (Cambridge University Press 2002) 186

⁹ Deirdre Madden, *Medicine, Ethics, and the Law*, (2nd edn, Bloomsbury Professional 2011) 45

¹⁰ *Devi v. West Midlands AHA* (1980) 7 CL 44

¹¹ *Re C (Adult: Refusal of medical treatment)* [1994] 1 WLR 290; [1994] 1 All ER 819

The classic legal conception of patient self- determination was provided by Justice Cardozo who stated “*Every human being of adult years and sound mind has the right to determine what shall be done to his own body.*”¹² In accordance with the right of autonomy and the right of self- determination, an individual therefore has a right to consent to or refuse medical treatment.

In the context of this debate, an individual therefore has the right to know their genetic make up and decide to undergo genetic testing, or ascertain their family history of illness. Individuals also have the right not to know. Indeed both the right to know and the right to not know one’s genetic make- up might be deemed to be an expression of one’s autonomy. Therefore, it can be asserted that the principle of autonomy provides a theoretical basis for such rights.¹³ The following section will further explore the right to know and the right not to know.

2.1 A right to know one’s genetic information

Individuals have a right to know their genetic make- up with a view to obtaining as much knowledge about themselves and their present and future health status. In cases where a disorder is potentially fatal but curable or preventable if diagnosed at an early stage, there are clear reasons why an individual would want to know their genetic make- up. Assuming that individuals want to live long and healthy lives, it would seem prudent to know as much as possible about potential predisposition to disease or disability. Knowledge of one’s genetic information gives an individual the opportunity to make necessary lifestyle and diet alterations that might contribute towards a healthier future.

In certain cases, there may be the possibility of prophylactic treatment options in cases where genetic conditions can be prevented. For example, upon discovery of the gene for hemochromatosis, (a common genetic condition which causes excess iron storage), a simple treatment is available that can ensure normal life expectancy.¹⁴ Knowledge of one’s genetic information offers the opportunity to better plan health care and treatment. It is acknowledged that in many cases in which genetic disease is indicated by the test result, there is no cure. Nevertheless, it is observed that “*even in the absence of therapies or cures, preparedness is often cited as a reason to seek out genetic knowledge.*”¹⁵ For financial, emotional or psychological reasons, there are strong reasons compelling discovery of one’s genetic make up. For example, couples interested

¹² *Schloendorff v. Society of New York Hospitals* 211 N.Y. 125 (1914)

¹³ Carol Lee, ‘Creating a Genetic Underclass: The Potential for Genetic Discrimination by the Health Insurance Industry’ (1993) 13 *Pace Law Review* 1 189, 205

¹⁴ For further details, see <http://www.hemochromatosisdna.com/> (accessed 6 February 2013)

¹⁵ Graeme T. Laurie, ‘Challenging Medical- Legal Norms: The Role of Autonomy, Confidentiality, and Privacy in Protecting Individual and Familial Group Rights in Genetic Information’ (2001) 22 *The Journal of Legal Medicine* 1

in starting a family may wish to know what genetic risks they may potentially pass on to their children, with a view to being prepared.¹⁶

In light of the personal nature of genetic information and because of the potentially harmful consequences if misused, individuals have an interest in controlling access to and use of their genetic information, incorporating a right to privacy, as well as a right not to be discriminated against. The right to know one's genetic make-up might be impacted by the fear of discrimination and breach of privacy. Individuals may be reluctant to engage in genetic testing and take advantage of genetic technologies out of fear of being denied employment or discriminated against in employment, as well as out of fear of being unable to access insurance, or other social goods and services.¹⁷ These issues also raise consideration of the right not to know.

2.2 The right to genetic ignorance

It might be assumed that enhanced knowledge is preferable and that knowing one's genetic make up is advantageous, on the assumption that most individuals want to live healthy and long lives. However, in cases where genetic conditions can be detected, but there is not yet a cure or effective treatment, for example, Huntington's disease, there are strong reasons why an individual might wish not to know their genetic make-up. A positive test result might provoke serious psychological consequences and have an adverse impact on an individual's mental health and well-being.¹⁸

A person's sense of self may be affected by the results of a genetic test and this may result in negative feelings about one's future. In this regard, it has been noted that "*the revelation of genetic hazard has been observed to result not only in repression but in anxiety, depression, and a sense of stigmatization.*"¹⁹ An individual might also be embarrassed by the potential stigma attached to having a predisposition to a genetic condition. Individuals who are reluctant to find out their genetic make up may be apprehensive of being presented with "*unwanted self-awareness.*"²⁰ In these circumstances, individuals may choose to remain ignorant of genetic status out of fear of revealing an incurable or untreatable

¹⁶ Deirdre Madden, *Medicine, Ethics and the Law* (2nd Ed, Bloomsbury Professional 2011) 306

¹⁷ Michael R. Santiago, 'Preventing Employment Discrimination Based on One's Genetic Characteristics' (1999) 30 McGeorge Law Review 703, 705

¹⁸ Lori B. Andrews, 'Legal Aspects of Genetic Information' (1990) 64 Yale Journal of Biological Medicine 29, 38. See also Elisabeth W. Almqvist, 'A worldwide assessment of the frequency of suicide, suicide attempts, or psychiatric hospitalization after predictive testing for Huntington's disease' (1999) 64 American Journal of Human Genetics 5, 1293. See also Michael R. Hayden, 'Predictive Testing for Huntington's Disease: Are we ready for widespread community implementation?' (1991) 40 American Journal of Medical Genetics 515

¹⁹ Daniel Kevles, *In the Name of Eugenics: Genetics and the Uses of Human Heredity* (Harvard University Press 1985) 298

²⁰ Colin S. Diver and Jane Maslow Cohen 'Genophobia: What is Wrong with Genetic Discrimination?' (2000-2001) 149 University of Pennsylvania Law Review 1440, 1442

condition.²¹ This may result in fear and produce negative psychological implications. An individual's psychological and mental health may be damaged by such disclosure of genetic information. In this context, Rabino refers to the emotional price of genetic testing.²² This right not to know is therefore "*designed to protect all those who prefer not to be haunted by the prospect of a predisposition to grave or even fatal biological conditions*,"²³ in circumstances where a genetic condition cannot be prevented.

The right not to know (and the right to know) has been recognised by various legal instruments. For example, the right not to know has been recognised in both the Council of Europe Convention on Human Rights and Biomedicine²⁴ and the UNESCO Universal Declaration on the Human Genome and Human Rights.²⁵ The international documents relating to these issues are discussed in chapter 6.

As highlighted, the right not to know also stems from the ethical principles of autonomy and self-determination. In accordance with these rights, an individual has a right to consent to or refuse medical treatment. An individual may therefore have the right not to know their genetic make-up and might choose not to avail of genetic testing. Indeed the right not to know one's genetic make-up might be deemed to be making an autonomous choice. Therefore the principle of autonomy provides a theoretical basis for a right not to know one's genetic make up.

Privacy is also referred to as a basis for the right not to know. It is submitted that "*true control of information must include the choice not to accept the information into one's private sphere*," and it has been observed that the "*imposition of unwarranted information*" can also be seen as a breach of privacy.²⁶ One commentator has noted that "[p]rivacy may be violated not only by the intrusion of a stranger, but by the compelling or persuading a person to direct too much attention to his own feelings and to attach too much importance to their analysis."²⁷ The right not to know is therefore firmly grounded in the right to privacy and confidentiality, and the recognition that health information, including genetic information is personal, and merits safeguarding. However, there are strong arguments against the right not to know which must be highlighted.

²¹ See Paul R. Billing *et al*, 'Discrimination as a Consequence of Genetic Testing' (1992) 50 American Journal of Human Genetics 476, 481. See also Sandi Wiggins *et al*, 'The Psychological Consequences of Predictive Testing for Huntington's Disease' (1992) 327 New England Journal of Medicine 1401

²² Isaac Rabino, 'Genetic Testing and its Implications: Human Genetics Researchers Grapple with Ethical Issues' (2003) 28 Science, Technology and Human Values 2, 365

²³ Noelle Lenoir, 'Universal Declaration on the Human Genome and Human Rights: The First Legal and Ethical Framework at the Global Level' (1998- 1999) 30 Columbia Human Rights Law Review 546, 564

²⁴ Council of Europe Convention on Human Rights and Biomedicine, Article 10(2)

²⁵ UNESCO Universal Declaration on the Human Genome and Human Rights, Article 5(c)

²⁶ Graeme Laurie, 'The Most Personal Information of All: An Appraisal of Genetic Privacy in the Shadow of the Human Genome Project' (1996) 10 International Journal of Law, Policy and the Family 74, 90

²⁷ James Fitzjames Stephen, *Liberty, Equality and Fraternity* (Henry Hold & Co 1873) 160

At this point, it can be observed that the interests of individuals might come into conflict with the potential competing rights of family members, and third parties. This is evident in circumstances where an individual chooses not to know their genetic information. The arguments against the right not to know are discussed in the following section.

2.3 Opposition to the right not to know

The right not to know may be criticised as being in contradiction with a patient's autonomy, with a physician's duty to inform patients, and with solidarity with family members, as well as on the grounds that it is irrational. The first argument is based on the view that foolishness should not be allowed.²⁸ This argument might be advocated on the part of the medical community when exercising the Hippocratic oath, and acting pursuant to principles of beneficence and non-maleficence. However, in light of the potential of serious psychological harm that might arise as a result of taking a genetic test, it could be argued that members of the medical profession have a responsibility not to inflict harm intentionally on a patient. In addition, the significance of beneficence is undermined to a certain extent by the emphasis on the importance of autonomy and the right to refuse medical treatment or information.²⁹

Linked to this, there is an argument that information is always good and it is therefore not advisable to remain in ignorance.³⁰ Such knowledge might also operate to enhance an individual's autonomy, and freedom to make informed choices regarding healthcare. Knowledge of a predisposition can provide individuals with the opportunity to change aspects of their lifestyle. However, it is also arguable that "*people are allowed to be foolish in liberal societies*,"³¹ and foolishness is not a reason to deny the freedom to act. This point might also be negated by an individual's right to make autonomous decisions.

In addition, there are arguments against the right to ignorance on the grounds of harm to others. Since genetic information about an individual also constitutes information about one's family members there might be circumstances where the ignorance of one person might cause harm to others.³² This argument acknowledges the link between families and genetic heritage. In light of the familial nature of genetic information, an individual who chooses not to know his or her genetic status "*could be said to be acting against solidarity*."³³ This is a

²⁸ Tuija Takala, 'The Right to Genetic Ignorance Confirmed' (1999) 13 Bioethics 3-4 288

²⁹ Deirdre Madden *Medicine, Ethics, and the Law*, (2nd edn, Bloomsbury Professional 2011) 46

³⁰ Aristotle stated that "*all men by nature desire to know*" – see Aristotle, *Metaphysics* (Oxford: Clarendon Press 1958)

³¹ *Re C (Adult, refusal of treatment)* [1994] 1 WLR 290; [1994] 1 All ER 819

³² Tuija Takala, 'The Right to Genetic Ignorance Confirmed' (1999) 13 Bioethics 3-4 288

³³ Roberto Andorno, 'The right not to know: an autonomy based approach' (2004) 30 Journal of Medical Ethics 435, 436

particularly strong argument when there is a potential of harm to the patient's family members who could be denied preventative measures.³⁴ Genetic test results may give family members the opportunity to make important lifestyle changes, with the objective of preventing or preparing for a potential disease.³⁵ These circumstances may give rise to a conflict of interests.³⁶

In addition, spouses and partners may have an interest in knowing genetic information. It has been argued that "*special duties arise when people make a commitment to one other such as in an intimate relationship, marriage or creating children.*"³⁷ These commitments create moral responsibilities to disclose genetic information.³⁸

2.4 Evaluation of an individual's rights

An individual has a direct interest in his/her genetic information, provoking consideration of the right to know and the right not to know. The theoretical basis of this interest stems primarily from the right to autonomy and self-determination. The right to know and the right not to know potentially clashes with other interests in genetic information, such as the family and third parties, creating a conflict of competing rights.

3. Family rights to genetic information

As highlighted, the individual's right to know draws attention to the familial nature of genetic information and highlights the potentially irreconcilable conflict of rights. This section will explore the issue of family rights. A potential conflict arises when an individual chooses not to know their genetic make up. This might create particular problems in circumstances where the lack of access to such information might potentially cause harm to family members. Disclosure of genetic information to an individual's relatives is premised on the same rationale as disclosure to an individual.

It is observed that a shift is emerging away from individual autonomy towards a more holistic approach, "*taking account not only of rights but of corresponding duties to others, particularly family members.*"³⁹ Accordingly, there is an inherent interconnectedness of rights and interests as regards genetic information. This merging of interests has led to the suggestion that the patient should be seen as the family rather than the individual.⁴⁰

³⁴ Ibid at 435

³⁵ Ibid at 437

³⁶ Ruth Chadwick, 'Genetics, choice and responsibility' (1999) 1 Health, Risk and Society 3 293

³⁷ Ann Somerville and Veronica English, 'Genetic Privacy: orthodoxy or oxymoron?' (1999) 25 Journal of Medical Ethics 144, 148

³⁸ Ibid

³⁹ Ibid at 145

⁴⁰ Ibid at 149

It is also submitted that this merging of interests has implications for the duty of confidentiality. Advances in genetics may force us to re- conceptualise the notion of medical confidentiality.⁴¹ In the context of confidentiality, the duty imposed on the doctor is not an absolute one and certain exceptions can be recognised. The doctor may be of the opinion that breach of this duty is justifiable in the interests of specific individuals who are affected by the information. In these circumstances, the rights of family members might be recognised, and a doctor might feel a responsibility to share details of familial genetic information. In this regard, it has been observed that *“the existence of a cure or treatment for the condition, the likelihood of harm to relatives, and the degree of severity if the condition does occur, are all relevant factors that would need to be considered.”*⁴² There may also be additional considerations if an individual is married or has a partner, particularly when there are reproductive choices at stake.

It is also important to acknowledge that the relative may indeed not want to know. In these circumstances, an individual may have decided to undergo genetic testing, whereas relatives may not want to know any details of their genetic status. In the same way that family members arguably have a right to know genetic information, they similarly have a right not to know, creating a further tension between the competing rights. Although they may be impossible to reconcile, the existence of these competing rights needs to be acknowledged.

3.1 Evaluation of family rights

One of the unique aspects of genetic information is its familial nature. This produces a complex range of ethical issues and competing interests, in ascertaining who the *“patient”* is, the duties of medical professionals, as well as the extent to which family rights to genetic information might take precedence over individual rights and vice versa. Although the familial and group nature of genetic information is noted, this thesis does not seek to address or reconcile this ethical dilemma. From a regulatory perspective, the focus of this thesis is on individual rights. It is however acknowledged that there might be a conflict with third party rights. The misuse of genetic information by third parties might impact upon the rights of the both individual and family members. The following section will discuss this aspect of the debate, in the employment and the insurance contexts.

4. Third party rights to genetic information

⁴¹ *Thomas Marshall (Exports) Ltd. V. Guinle* [1979] 3 AL ER 193

⁴² For further discussion see Graeme T. Laurie, ‘The most personal information of all: An appraisal of genetic privacy in the shadow of the human genome project’ (1996) 10 *International Journal of Law, Policy and the Family* 74, 85

Genetic information is also attractive to third parties and may be viewed as a powerful predictive tool, primarily for economic reasons. In highlighting the commercial interests at stake, the right to know must therefore be explored within the context of third parties and the extent to which such third parties might legitimately access and use genetic information. There are many third party contexts in which genetic information might be of interest, for example, in employment, insurance, commercial lending, education, sport, immigration and reproduction. However, for the purposes of this thesis, the focus is narrowed to the employment and insurance contexts.

It is noted that employment and insurance are both portals to accessing a myriad of social and economic goods and services, thereby impacting upon an individual's life and participation in society. It is important to note that although genetic testing by employers and insurers is not yet taking place on a widespread basis, rapidly advancing genetic technology indicates that testing will undoubtedly become cheaper,⁴³ more sophisticated and therefore more accessible. Indeed it has been acknowledged that advances in genetics have led many to hypothesise that widespread genetic discrimination is to be anticipated.⁴⁴

This section will illustrate how third party rights result in a conflict with individual and family rights, as well as with broader societal interests. Specifically, third party interest in genetic information and the potential right to know may result in breach of privacy and discrimination, and ultimately create a society classified according to genetic desirability. Discussion of third party interests in genetic information also highlights the societal interest in genetic science and the appropriate application of genetic science.

Before exploring the use of genetic information in employment and insurance, the next section will explain the key concepts of genetic discrimination and genetic privacy.

4.1 Genetic discrimination

This section will introduce the concept of genetic discrimination. Discrimination involves making distinctions between individuals on the basis of certain characteristics, such as age, race, disability or sex and using those distinctions as the basis for differential treatment.⁴⁵ The increasing use of genetic testing and the rise in the availability of genetic information have "*unveiled a new dimension*

⁴³ John Markoff, 'Cost of Gene Sequencing Falls, Raising Hopes for Medical Advances' *New York Times* (New York 7 March 2012)

⁴⁴ Jennifer Chorpene, 'Genetic Disability: A Modest Proposal to Modify the ADA to Protect Against some forms of Genetic Discrimination' (2004) 82 *North Carolina Law Review* 1441, 1443. See also Pauline T. Kim, 'Genetic Discrimination, Genetic Privacy: Rethinking Employee Protections for a Brave New Workplace' (2002) 96 *Northwestern University Law Review* 1497, 1498

⁴⁵ The theory of non-discrimination will be further discussed in chapter 5

or layer of human difference, that of genetic makeup,”⁴⁶ which offers a new means by which to differentiate between individuals.

The term genetic discrimination has been explored extensively in literature. Genetic discrimination can refer to the differential treatment of individuals or their family on the basis of an apparent, or assumed variation from “*normal*” human genes. In general terms, it can be described as a form of differentiation based on genetic features or based on genetic information. It has also been described as “*discrimination against an individual or against members of that individual’s family solely because of real or perceived differences from the ‘normal’ genome in the genetic constitution of that individual.*”⁴⁷ Rothstein has used the term to describe the differential treatment based on genetic status.⁴⁸ The common thread identified is differential or discriminatory treatment on the basis of genetic information. Genetic discrimination has the potential of creating “*a new group of disadvantaged people...*”⁴⁹

As noted in chapter 2, genetic information can be obtained, not only from genetic test results, but also from other sources, such as family history. Therefore, for the purposes of this thesis, it is acknowledged that discrimination can occur on the basis, not only of genetic test results, but also on the basis of family history information.⁵⁰ Therefore, the discriminatory use of genetic information gleaned from family history (and other potential sources) comes within the scope of the concept of genetic discrimination.

A key element of the concept of genetic discrimination is that it describes discrimination against asymptomatic individuals, in other words, those who have not yet expressed symptoms of disease or disability. Natowicz *et al* “*distinguish genetic discrimination from discrimination based on disabilities caused by altered genes,*” and thereby clarify that genetic discrimination is based on information inferred from an individual’s genotype, as opposed to on the basis the current symptoms (or phenotype) of an individual.⁵¹ A similar point is made by Yesley who notes that “*the quintessential feature of genetic discrimination is the use of genetic information about an asymptomatic person.*” It is noted that if a genetic

⁴⁶ Anne Mainsbridge, ‘Employers and Genetic Information: A New Frontier in Discrimination’ (2002) 2 Macquarie Law Journal 61, 67

⁴⁷ Marvin R. Natowicz *et al*, ‘Genetic Discrimination and the Law’ (1992) 50 American Journal of Human Genetics 465, 466. A similar definition has been given by Paul R. Billings *et al*, ‘Discrimination as a Consequence of Genetic Testing’ (1992) 50 American Journal of Human Genetics 476, 476

⁴⁸ Mark A. Rothstein, ‘Genetic Discrimination in Employment: Ethics, Policy and Comparative Law’, in Swiss Institute of Comparative Law (ed), *Human Genetic Analysis and the Protection of Personality and Privacy* (Schulthess Polygraphischer Verlag 1994) 129

⁴⁹ Marvin R. Natowicz *et al*, ‘Genetic Discrimination and the Law’ (1992) 50 American Journal of Human Genetics 465, 465

⁵⁰ Margaret Otlowski *et al*, ‘Practices and Attitudes of Australian Employers in relation to the use of genetic information: Report on a National Study’ (2009- 2010) 31 Comparative Labor Law and Policy Journal 637, 638

⁵¹ *Ibid.*

condition expresses itself, it can be classified as another type of discrimination, namely discrimination on the basis of one's health or disability.⁵²

From a disability perspective, genetic testing can predict a propensity to or probability of future disability (as explained in chapter 2). Genetic discrimination can therefore be described as discrimination on the basis of potential future disability, or phrased differently, on the basis of genetic predisposition to disability.⁵³ Similarly, genetic discrimination can signal negative attitudes towards disability and genetic conditions, and the concept may also encompass assumed, perceived or imputed disability. On the basis of a family history of illness or following a positive genetic test result, it might be assumed either that a person has that illness or disability, or will certainly develop such a genetic condition. Accordingly, if genetic information is too widely available, it exposes putative persons with disabilities to overt and covert discrimination.

Genetic discrimination as a concept can be described to mean, not only differential treatment, but also, in certain contexts (for example, in the insurance industry), unfair differential treatment, which cannot be objectively justified. It is also necessary to point out that every individual is a potential victim of genetic discrimination, as everyone carries genes that predispose to a variety of common illnesses.⁵⁴

4.2 Genetic privacy

In addition to discrimination, the right to privacy is a concern.⁵⁵ As explained, one of the unique features of genetic information is that it reveals sensitive, personal information about not just an individual, but that individual's family.⁵⁶ Therefore, there are compelling reasons why an individual and an individual's family would wish for genetic information to remain private and confidential, and safe from outside intrusion. In light of the sensitive nature of genetic information, the potential for abuse is clear, as is the desire to protect such information from access and disclosure. It is noted that an individual's (and family members') privacy may be breached in circumstances where third parties gain access to or

⁵² Michael S. Yesley, 'Protecting Genetic Difference' (1999) 13 Berkeley Technology Law Review 653, 662

⁵³ See generally, Paul S. Miller, 'Genetic Testing and the Future of Disability Insurance: Thinking about Discrimination in the Genetic Age' (2007) Journal of Law, Medicine and Ethics 47. See also Gregor Wolbring, 'A Disability Rights Approach to Genetic Discrimination', in Judit Sandor (ed) *Society and Genetic Information: Codes and Laws in the Genetic Era* (Central European University Press 2004) 161

⁵⁴ See comments by Dr Francis Collins, Former Director, National Human Genome Research Institute, Regarding the Passage of Genetic Information Nondiscrimination Act of 2005 (S. 306) (17 February 2005), <http://www.genome.gov/13014311> (accessed 16 April 2012)

⁵⁵ For further discussion see Mark Taylor, *Genetic Data and the Law: A Critical Perspective on Privacy Protection* (Cambridge University Press 2012)

⁵⁶ Graeme Laurie, 'The Most Personal Information of All: An Appraisal of Genetic Privacy in the Shadow of the Human Genome Project' (1996) 10 International Journal of Law, Policy and Family 74-101

use genetic information without the appropriate consent. As a fundamental right, all individuals are entitled to protection of their privacy.⁵⁷

It is observed that disclosure of medical information in the context of employment leaves the employee particularly vulnerable, because employers are interested in this information, partly to make decisions that could adversely affect the employee, both at the pre- employment stage and also throughout the course of employment. Therefore, there is a strong argument for privacy in these relationships. Similarly, in the insurance context, disclosure of genetic information can result in the insured being particularly vulnerable because the insurer can make decisions based on such information, which may affect the insurability of the individual.

The concept of privacy also attracts consideration of personhood, particularly in the context of discussing genetic information, thereby enhancing the argument in favour of protecting genetic privacy. Privacy protects the integrity of self and *“allows one to limit access to oneself and to avoid being the object of scrutiny, which gives us space for maintenance and development of the self, for the quest that shapes the self.”*⁵⁸ The absence of privacy has negative implications on concepts of personhood and self- awareness.⁵⁹ Privacy therefore protects information that is integral to the self, such as genetic information. In consideration of the personal nature of genetic information, it has also been noted that genetic privacy *“protects personhood interests in shielding us against stigmatization, discrimination and being misunderstood.”*⁶⁰ The desire to protect core concepts of personhood compounds the need to reflect upon the notion of genetic privacy, and ensure that privacy is a key consideration in this debate. The concepts of privacy and non- discrimination, as well as their rationale and sources in law will be further discussed in chapter 5.

Having highlighted the concepts of genetic discrimination and genetic privacy, which are the main concerns in the third party context, the next section will explore in further detail the use of genetic information in employment and insurance.

5. Use of genetic information in employment

It is noted that employers may have an interest in genetic information derived from an individual's family history of illness, as well as genetic information obtained from the results of genetic tests previously taken. Employers may also

⁵⁷ For example, European Convention on Human Rights, Article 8

⁵⁸ Sonia Suter, 'Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy' (2003- 2004) 72 The George Washington Law Review 737, 771

⁵⁹ See generally Jeffrey H. Reiman, 'Privacy, Intimacy, and Personhood' (1976) 6 Philosophy and Public Affairs 1 26

⁶⁰ Sonia Suter, 'Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy' (2003- 2004) 72 The George Washington Law Review 737, 776

have an interest in requesting that employees undergo genetic testing. In ascertaining the basis for an employer's interest in genetic information, it is observed that employers first of all have an obvious financial interest in using genetic information. It is also acknowledged that they have a concern for protecting the employee and the prospective employee.⁶¹ On the basis of this interest, an employer may cite health and safety reasons for wishing to use genetic information.

Genetic testing in the workplace generally incorporates the practices of genetic screening and genetic monitoring, as discussed in chapter 2. As outlined in previous chapters, the practice of genetic testing can offer valuable insights into an individual's current health status, as well as predisposition to illness, thereby offering insights into potential future health status, illness and disability. The following section will explain the factors contributing to an employer's interest in genetics, from both an economic perspective, and from a health and safety perspective.

5.1 The preference for healthy and productive workers

Employers generally endeavour to maximise employees' fitness and productivity while minimising turnover and employee absenteeism.⁶² When employees are in ill health, they are more likely to require sick pay, are more likely to suffer occupational injury or illness, and their illnesses may impact upon the morale of co-workers and other third parties.⁶³ Further, when an employee becomes ill or disabled while in employment, productivity may suffer, and future productivity is uncertain. The costs and time of hiring and training replacement employees may be significant, with adverse implications for efficiency. Ill health may also result in retirement or redundancy, requiring financial payouts to employees. These factors collectively result in increased costs and overheads for the employer.

When hiring, training or promoting employees, employers therefore have strong economic incentives to select a desirable candidate who will yield a favourable return on their investment. Hiring or promoting an under qualified and under productive worker is costly to the employer. In addition, employers who facilitate health insurance for employees have increased economic incentives to be concerned about the productivity of workers. Therefore the ill health or potential ill health of employees can have numerous adverse financial consequences for employers.

⁶¹ Margaret Otlowski *et al*, 'Practices and Attitudes of Australian Employers in relation to the use of genetic information: Report on a National Study' (2009- 2010) 31 *Comparative Labor Law and Policy Journal* 637, 640

⁶² Mark A. Rothstein *et al*, 'Protecting Genetic Privacy by Permitting Employer Access Only to Job-Related Employee Medical Information: Analysis of a Unique Minnesota Law' (1998) 24 *American Journal of Law and Medicine* 399, 401

⁶³ Mark. A. Rothstein and Bartha Knoppers, 'Legal Aspects of Genetics, Work and Insurance in North America and Europe' (1996) 3 *European Journal of Health Law* 143, 144

In light of the fact that employers clearly have an interest in minimising employment costs, “*the supposedly predictive nature of genetic information is a very attractive tool in workplace management.*”⁶⁴ Healthy employees who are perceived to possess desirable genes arguably do not create these financial problems. Use of genetic information may potentially improve the productivity of current as well as potential employees. Indeed, obtaining genetic information about employees and potential employees may be viewed as sensible commercial practice.⁶⁵ Employers may seek to take advantage of advancing genetic technology by requiring the use of genetic testing as a prerequisite to employment, or by requesting existing genetic test results or details of family medical history. By selecting genetically desirable employees, employers may seek to employ only those who will remain healthy and productive and those who are least likely to be a liability.⁶⁶ Similarly, genetic information might also be of interest to employers when making termination decisions, particularly in recessionary times.

At a more general level, it may be further argued that an employer has a legitimate interest in obtaining as much information as possible about a prospective employee in order to maximise its prospects of choosing the most appropriate possible person for the job.⁶⁷ Advancing genetic science can uncover potentially useful profiles – for example, the prevalence of the “*risk-taking*” gene,⁶⁸ which some employers may view as desirable in a potential employee, while others may see this as indicative of a high-risk candidate. As genetics becomes more closely connected to behavioural and personality traits, employers have additional incentives to take advantage of new technologies to find a candidate who fits the profile of their organisation.

⁶⁴ Graeme Laurie, *Genetic Privacy – A Challenge to Medico- Legal Norms* (Cambridge University Press 2002) 153

⁶⁵ Susannah Carr, ‘Invisible Actors: Genetic Testing and Genetic Discrimination in the Workplace’ (2007- 2008) 30 University of Arkansas at Little Rock Law Review 2, 2

⁶⁶ Margaret Otlowski *et al*, ‘Practices and Attitudes of Australian Employers in relation to the use of genetic information: Report on a National Study’ (2009- 2010) 31 Comparative Labor Law and Policy Journal 637, 640

⁶⁷ Anne Mainsbridge, ‘Employers and Genetic Information: A New Frontier in Discrimination’ (2002) 2 Macquarie Law Journal 61, 66

⁶⁸ Richard Alleyne ‘Why risk taking runs in the family – scientists find gene that makes you gamble’, *The Telegraph* (London 6 May 2009) <http://www.telegraph.co.uk/news/newstopics/howaboutthat/5279222/Why-risk-taking-runs-in-the-family-scientists-find-gene-that-makes-you-gamble.html> (accessed 7 June 2012). See also Scott Shane, ‘Explaining Innate Risk- Takers’, *Bloomberg Businessweek* (12 February 2010). Available at: http://www.businessweek.com/smallbiz/content/feb2010/sb20100211_567240.htm (accessed 7th June 2012), citing Scott Shane, *Born Entrepreneurs, Born Leaders – How your Genes Affect your Work Life* (Oxford University Press 2010). See also Mary J. Kreek *et al*, ‘Genetic Influences on impulsivity, risk- taking, stress, responsibility and vulnerability to drug abuse and addiction’ (2005) 8 Nature Neuroscience 11 1450

In this context, it is noted that genetic testing could be viewed as an extension of medical testing, which has traditionally been a standard employment practice.⁶⁹ An employer is generally concerned with selecting the right physical and psychological profile for a given job. The employer may wish to ask questions or perform various tests to confirm whether candidates are suitable for employment or whether they would pose a risk to health or safety. The question then arises whether these assessments are “*unnecessary or overly intrusive*.”⁷⁰ It is noted that where such medical questions are too personal, “*the focus will inevitably be upon the necessity or justification for such intrusions where they occur*.”⁷¹ In applying the ethos of this standard employment practice to the practice of genetic testing, there may be scope to argue that from the employer’s perspective, genetic testing is an extension of medical testing in terms of its objective in ascertaining the suitability of the potential employee. The question then arises as to the necessity or justification for such testing. This falls upon consideration of an employer’s right to know and the limits that should be placed on that right, which will be expanded upon throughout this section.

In the context of examining the employer’s interest in genetic information, it is established that the employer has clear economic reasons for seeking access to and use of employee’s genetic information. In certain circumstances this interest may be viewed as an extension of the practice of medical testing. The following section will examine a further reason behind an employer’s interest in genetic information.

5.2 Use of genetics to address occupational disease

An employer’s wish to obtain genetic information may be motivated by occupational health and safety considerations.⁷² For example, an employer may wish to use genetic information to determine whether or not an employee has a genetic predisposition that may pose a safety risk to the employee, co-workers or the public or that may cause the employee to be particularly sensitive to workplace hazards.⁷³ From a health and safety perspective, medical questions and analysis, potentially encompassing genetic testing, may be required to protect against hazards at work and to fulfill an employer’s obligations in this regard. The following section will highlight the applicable law in this area.

⁶⁹ See generally Aart Hendriks and Sjef Gevers, ‘(Pre-) employment Medical Examinations and the Law, with Particular Reference to the European Union’ (1994) 1 *European Journal of Health Law* 229

⁷⁰ Cathal McGreal, ‘Workplace Privacy and Data Protection’ in Maeve Regan (ed) *Employment Law* (Tottel Publishing 2009) 212

⁷¹ *Ibid* at 212

⁷² Susannah Carr, ‘Invisible Actors: Genetic Testing and Genetic Discrimination in the Workplace’ (2007- 2008) 30 *University of Arkansas Law Review* 1

⁷³ Margaret Otlowski *et al*, ‘Practices and Attitudes of Australian Employers in relation to the use of genetic information: Report on a National Study’ (2009- 2010) 31 *Comparative Labor Law and Policy Journal* 637, 640

5.3 Health and safety obligations

An employer has obligations under health and safety law, both under statute⁷⁴ and under common law. Pursuant to health and safety law, various duties and powers now provide for medical questions, testing and assessments in employment. The common law duty of care towards the employee also imposes certain duties on employers to avoid liability in tort law.⁷⁵

In light of an employer's obligations under health and safety law, genetic monitoring could potentially be used to screen job applicants and employees, who, because of their genetic status, may be at an increased risk of developing certain diseases if exposed to certain occupational chemicals or other hazards.⁷⁶ Individuals with the sickle cell trait, for example, may be at increased risk for sickle cell anaemia if exposed to carbon monoxide or cyanide. Similarly, carpal tunnel syndrome may also be exacerbated in certain work environments, particularly in office environments in which employees use computers on a frequent basis, or where an employee is engaged in a significant amount of repetitive work.⁷⁷ Surveillance is one way to monitor carpal tunnel syndrome in the workplace. In carrying out such surveillance, an employer may wish to carry out a genetic test to ascertain those who might be predisposed to developing this condition.

It is also acknowledged that employees have corresponding duties to take care of their own health and safety in the workplace. In the context of this discussion, knowledge of one's genetic make-up may trigger action on the part of an employee to take a proactive role in relation to workplace health and safety. Arguably, it may also potentially act as a defence to employer's liability in certain circumstances.

5.4 Evaluation of use of genetic technologies for health and safety reasons

Use of genetic technology can be viewed as a potentially effective mechanism for reducing the incidence of occupational disease, both for the benefit of the

⁷⁴ Council Directive 89/391/EEC of 12 June 1989 on the introduction of measures to encourage improvements in the safety and health of workers at work [1989] OJ L 183/1

⁷⁵ *Dalton v Frendo*, Supreme Court, 15 December 1997, p 4 of O'Higgins CJ's judgment; p 5 of Griffin J's judgment. See also *Wilsons and Clyde Coal Co. Ltd v English* [1938] AC 57, House of Lords. As regards the obligation to provide a safe place of work, see *Gallagher v Mogul of Ireland Ltd* [1975] IR 204 (SC)

⁷⁶ Melinda B. Kaufman, 'Genetic Discrimination in the Workplace: An Overview of Existing Protections' (1998- 1999) 30 Loyola University Chicago Law Journal 393

⁷⁷ Keith T. Palmer *et al*, 'Carpal Tunnel Syndrome and its relation to occupation: a systemic literature review' (2007) 57 Occupational Medicine 57. See also, Nancy Gell *et al*, 'A Longitudinal Study of Industrial and Clerical Workers: Incidence of Carpal Tunnel Syndrome and Assessment of Risk Factors' (2005) 15 Journal of Occupational Rehabilitation 1 47

employer, the employee and the public.⁷⁸ The employer has an interest in avoiding liability for any workplace accident or illness, and in maintaining their duty of care to employees. Employees arguably have an interest in obtaining information about their genetic status in hazardous workplaces in order to monitor their health, safety and welfare. By reducing occupational disease and injury, genetic monitoring may minimise employment costs.⁷⁹ Accordingly, there are also economic motives behind use of such genetic monitoring.

Although there are strong arguments in favour of an employer's use of genetic monitoring in hazardous workplaces, caution must be exercised to ensure that employees' rights are maintained and certain safeguards must be put in place. For example, it is important to have regard to an employee's privacy and data protection rights,⁸⁰ and best practice would be to ensure that such monitoring is carried out on a voluntary basis, and with the employee's informed consent. It is also important to have regard to the limited accuracy and reliability of genetic technologies.

5.5 Consequences of misuse of genetic information in employment

This section will look at the undesirable implications of using genetic information from the perspective of the employee. Firstly, this section will look at the discriminatory use of genetic information by employers. Employers arguably have no right to require that an individual undergo genetic testing (or use the results of genetic tests previously taken) when that individual displays no indication of a disease or disability that would inhibit the current ability to do a job. Making an employment decision on the basis of the probability of an individual developing a certain disease or disability, as opposed to on the actual ability to perform the work, can be deemed to be unlawful discrimination. In the EU, there is strong legislation promoting equality in the workplace.⁸¹

Discrimination based upon actual or perceived genetic characteristics denies an individual equal opportunity on the grounds of a genetic status over which he/she has no control. Discrimination based on genetic information can be as unjust as that based on race or gender (or other similar characteristics). It is noted that *"the right to be treated equally and according to one's abilities in all the diverse*

⁷⁸ Janneke H. Gerards and Heleen L. Janssen, 'Regulation of Genetic and Other Health Information in a Comparative Perspective' (2006) 13 European Journal of Health Law 339, 363

⁷⁹ Robert A. Bohrer, 'A Rawlsian Approach to Solving the Problem of Genetic Discrimination in Toxic Workplaces' (2002) 39 San Diego Law Review 747. See also Michael Baram, 'Genetic Testing for Susceptibility to Disease from Exposure to Toxic Chemicals: Implications for Public and Worker Health Policies' (2000- 2001) 41 Jurimetrics 165

⁸⁰ Council and Parliament Directive 95/46/EC of 24 October 1995 on the protection of individuals with regard to the processing of personal data and on the free movement of such data [1995] OJ L 281/31

⁸¹ See chapter 9

*aspects of human endeavour is a core social value.*⁸² Genetic discrimination, as described above, therefore violates basic tenets of justice and is potentially harmful to societal values.

Similarly, the potential for indirect discrimination is also recognised. In this scenario, discrimination may be deemed to arise from employment actions, practices or provisions which are shown to have an adverse effect on an individual or group of individuals. For example, past employment practices, such as genetic screening for sickle cell anaemia in the US in the 1970s resulted in an adverse impact on African Americans (discussed below). Other types of genetic tests, such as those indicating susceptibility to breast cancer might also have a more adverse effect on women, than on men. There is a potential that employers might introduce a genetic screening programme that appears to target all individuals on an equal basis, but produces negative effects for certain individuals, or possibly racial or ethnic groups. Discrimination (both direct and indirect) on the basis of genetic information is a real concern, and one with potentially serious consequences for employees and potential employees.

In addition, misuse of sensitive genetic information can result in the violation of an employee's privacy. Unauthorised disclosure of genetic information to employers may breach the right to privacy, leaving individuals in a vulnerable position.⁸³ There is a strong societal interest in ensuring the protection of privacy, as evidenced by the strong data protection laws which are in place in the EU. Linked to this, doctor- patient confidentiality may be infringed if an employer seeks an employee's medical records.

In addition to concerns of discrimination and privacy, there are further ethical issues that arise, such as the right to know one's genetic information and the right not to know. These rights come into conflict with an employer's right to access and use genetic information. From a legal and ethical perspective, misuse of genetic information by employers therefore has undesirable consequences for the employee and that employee's family members. Such information can be used to discriminate against employees and potential employees at the hiring, promotion or termination stages, and may have adverse implications for one's privacy. It is observed that there are further societal consequences that may arise in the employment context, which will be discussed below.

5.6 Wider implications of misusing genetic information

⁸² Lawrence O. Gostin, 'Genetic Discrimination: The Use of Genetically Based Diagnostic and Prognostic Tests by Employers and Insurers' (1991) 17 American Journal of Law and Medicine 109, 112

⁸³ For further discussion see Mark Taylor, *Genetic Data and the Law: A Critical Perspective on Privacy Protection* (Cambridge University Press 2012)

Individuals who are refused employment on the grounds of genetic information are left without a job. By excluding otherwise suitable individuals from employment, society is drained of skills and innovation. This exclusion “*promotes physical and economic dependency, draining rather than enriching social institutions.*”⁸⁴ Consequently, genetic technology can create employment barriers by effectively classifying individuals by potential future health risk. Society may lose out on the contribution of these individuals, who are willing and in a position to make an active and productive addition to the community, and who are being denied this opportunity, despite their current ability. This may result in the further exclusion and isolation of vulnerable persons, such as persons with putative disabilities and the elderly, and create further barriers to their integration into the community.

Therefore, access to employment may well act as a gateway to and may impact upon access to other social and economic goods and services (including health care). It may impact upon fulfilling civic engagement, including access to political life and active involvement in the community. Access to employment may also potentially affect an individual’s social life, friendships and interaction with peers, which has a significant impact on inclusion and active participation in society. Therefore, at a fundamental level, the existence of barriers to employment by way of misusing genetic information may have a profound impact on an individual’s private and professional life.

5.7 Striking a balance

It has been asserted that genetic testing in the employment context is “*a technology both full of promise and fraught with ethical peril.*”⁸⁵ It raises considerations of privacy and discrimination. It can also potentially exacerbate existing racial and class inequalities, with negative implications for the individual and society. Just as it is unlawful to refuse to hire someone on the basis of race or gender,⁸⁶ it should be unlawful to refuse to hire an individual on the basis of genetic information (without considering that individual’s qualifications and current ability to do the job).⁸⁷ It is submitted that an otherwise suitable individual should not be prevented from obtaining or maintaining a job based upon having a certain genetic predisposition. Employers also have a responsibility to uphold employees’ privacy and uphold data protection laws.

⁸⁴ Lawrence O. Gostin, ‘Genetic Discrimination: The Use of Genetically Based Diagnostic and Prognostic Tests by Employers and Insurers’ (1991) 17 American Journal of Law and Medicine 109, 112- 113

⁸⁵ Chris MacDonald and Bryn Williams- Jones, ‘Ethics and Genetics: Susceptibility Testing in the Workplace’ (2002) 35 Journal of Business Ethics 235, 238

⁸⁶ See chapter 9 for discussion of the EU non- discrimination framework

⁸⁷ Paul Steven Miller, ‘Is there a pink slip in my genes? Genetic discrimination in the workplace’ (1999- 2000) 3 Journal of Health Care Law and Policy 2 225, 264

In addressing this area, it is important to consider the competing rights, and assess where the appropriate balance should be struck. As highlighted, there are strong arguments in favour of an employer's use of genetic technologies, primarily for health and safety reasons. However, controls need to be placed on this right, for example, having regard to the accuracy of genetic science. It is important that the limited accuracy of genetic testing be acknowledged, in considering any potential employer use of such technology. An employee's rights also need to be upheld. As explained, it is important to ensure protection against unfair discrimination in hiring, promotion and termination and to maintain employee privacy. To facilitate such rights, it is submitted that there must be safeguards limiting an employer's access to and use of genetic information.

It is also necessary that this balance include consideration of society's interest in reaping the benefits of advancing genetic science. This goal may be reached by ensuring that the area is appropriately regulated, taking account of the fundamental rights of employees, and the limited right to know of employers in certain workplace scenarios.

6. Use of genetic information in insurance

Genetic information is also raising concerns in the insurance industry. From the outset it is important to make some observations in relation to the insurance industry, the different systems of insurance and the different types of insurance products available. Firstly, there is a distinction made between the solidarity based system of insurance and the mutuality based system of insurance.⁸⁸ In solidarity- based systems, every individual is insured against a particular type of risk, without there being a clear correlation between an individual's risk and the premium paid.⁸⁹ In contrast, in mutuality- based systems, an accurate classification is made in risk pools, with each risk group being charged a premium that corresponds with the estimated insurance risk pool.⁹⁰

Most private insurance systems are based on mutuality and it is acknowledged that this debate sparks particular resonance in the realm of private insurance (where risk classification is a fundamental practice). It is also necessary to highlight the different types of private insurance relevant to this discussion, with the issue of genetic information being most immediately relevant in the case of health insurance and life insurance.⁹¹ Accordingly, this thesis will focus on health and life insurance in this context.

⁸⁸ Onora O'Neill, 'Insurance and Genetics: the Current State of Play' (1998) 61 *Modern Law Review* 571-6

⁸⁹ Yann Joly *et al*, 'Genetic discrimination in private insurance: global perspectives' (2010) 29 *New Genetics and Society* 251, 353

⁹⁰ *Ibid* at 354

⁹¹ James Mittra, 'Predictive genetic information and access to life insurance: the poverty of genetic exceptionalism' (2007) 2 *Biosocieties* 3-49

The societal and public view of life insurance can be compared with the view of health insurance. In most societies, health insurance and access to health care are universally viewed as a necessary and basic social good that should be available and accessible to all individuals. In most EU countries, there exists primarily publicly funded health care. On the other hand, in jurisdictions such as the US, it is primarily a private health care model. The individual is required to purchase their own private insurance. Accordingly, the social role of insurance differs greatly in different jurisdictions.

Discussion of the use of genetic information in the area of health insurance therefore generally varies according to the existence or not of a universal health care system. In jurisdictions without a universal health care system, there is a more obvious emphasis on the importance of private health insurance (such as the US). However, although most EU countries have some form of universal access to health care, private health insurance is still an important commodity, and a necessary and thriving social good (as is life insurance). Individuals generally purchase private health insurance with a view to obtaining additional services and ensuring faster access to health care. Indeed, legislative initiatives in this area have nevertheless taken place in countries with some form of universal access to health care. Having clarified the different systems insurance and the importance in society, the following section will discuss an insurer's interest in genetic information.

6.1 An insurer's interest in genetic information

Access to and use of genetic information by insurance companies raises concerns in relation to an individual's privacy, access to insurance (and health care), and the possibility of discrimination. As a profit-driven industry, there are strong economic reasons to justify the use of genetic information. Insurers may wish to use advancing genetic science for financial advantage and as a tool for selecting what they perceive to be low risk customers. From the perspective of the insurance company, it can be argued that the economic viability of the industry relies upon disclosure of all relevant information.

The following section will briefly highlight the fundamental principles of insurance law as a prelude to the impact of genetic information in insurance. This section will also illustrate how an insurer's economic interest in genetic information may conflict with an individual's right not to be discriminated against, right to privacy and society's interest in ensuring access to insurance and other social goods and services.

6.2 Fundamental principles of insurance law

On considering the impact of genetic information on the insurance industry, one of the primary considerations is the concept of *uberrimae fides*. In common law

jurisdictions, insurance law is premised upon the concept of utmost good faith or *uberrimae fides*,⁹² and provides that both parties to the insurance contract must disclose all relevant, material⁹³ information.⁹⁴ The duty of utmost good faith was explained in *Carter v Boehm*.⁹⁵ Insurance law generally requires insured persons to inform the insurer of all relevant facts known to them.⁹⁶ The principle of *uberrimae fides* has been well documented and it is widely established that this core principle forms a fundamental part of insurance law. Pursuant to the principle of *uberrimae fides*, any failure to disclose material facts will result in the contract being avoided by the insurer (*ab initio*), with the result that the contract is deemed never to have existed.⁹⁷ It might be argued that failure to disclose the results of a genetic test, or details of family history of illness may be contrary to the duty of utmost good faith.

6.3 The practice of risk assessment

This section will outline the basis upon which insurance companies assess risk and charge premiums. In order to predict claims, it must gather certain information regarding the characteristics of its insureds. Insurance companies use this particular information in a process called underwriting, the premise of which is that the insured should pay a premium according to the individual risk.⁹⁸ Underwriting has been defined as “*the process by which insurance companies classify individuals according to their predicted risk for the purpose of deciding whether to offer or refuse them coverage, what restrictions to impose on any coverage offered to them, and what rates to charge them for any coverage offered.*”⁹⁹ Underwriting has also been described as the method used to find “*the best and most desirable insureds.*”¹⁰⁰

⁹² *Bell v Lever Bros Ltd* [1932] AC 161, 227; *Banque Keyser SA v Skandia (UK) Ins Co Ltd* [1990] 1 QB 665, 769

⁹³ The test of materiality provides that, ‘Every circumstance is material which would influence the judgment of a prudent insurer in fixing the premium or determining whether he will take the risk.’ See *Lambert v Cooperative Insurance Society* [1975] 2 Lloyd’s Rep 485; *Pan Atlantic Insurance Company v Pine Top Insurance Co Ltd* [1995] 1 AC 501

⁹⁴ A leading case that established the duty of disclosure in insurance is *Carter v Boehm* (1766) 3 Burr. 1905. For further discussion of the doctrine of *uberrimae fides*, see Reuben A. Hasson, ‘The doctrine of *uberrimae fides* in insurance law: a critical evaluation’ (1969) 32 Modern Law Review 615

⁹⁵ *Carter v Boehm* (1766) 3 Burr 1905

⁹⁶ See generally, John Lowry and Philip Rawlings, *Insurance Law: Doctrines and Principles* (2nd edn, Hart Publishing 1999)

⁹⁷ *Ibid.*

⁹⁸ Roberta M. Berry, ‘The Human Genome Project and the End of Insurance’ (1996) 7 University of Florida Journal of Law and Public Policy, 2 205, 216

⁹⁹ *Ibid.*

¹⁰⁰ Deborah A. Stone, ‘The Struggle for the Soul of Health Insurance’ (1993) 18 Journal of Health Politics, Policy and Law 2 287, 294

In accordance with the concept of underwriting, risk assessment and classification of insured persons are central principles.¹⁰¹ Insurers have generally created classifications to recognise the differences that exist between individuals, encompassing both intrinsic factors and optional lifestyle choices. This allows insurance companies to employ “*reasonable discrimination*” and establish premiums on “*a particular individual’s projected costs*.”¹⁰² In this regard, it has been asserted that the use of the word “*discrimination*” with respect to insurance may be misleading, because insurance underwriting by its very nature involves treating people in different risk classes differently, and therefore “*inherently entails discrimination*.”¹⁰³ The objective is to ensure that each insured person’s premium is based on the amount of his or her risk.¹⁰⁴

In the context of health and life insurance, there are a number of factors which are generally necessary to calculate risk. The factors that impact upon risk classification (such as gender, age, health, occupation, lifestyle choices for example, smoking), are analysed by insurance companies in assessing premiums. Some of these factors are immutable and intrinsic, such as age, gender and an individual’s genetic information, while other factors, such as smoking and drinking are optional lifestyle choices. Different rates based on the optional lifestyle factors seem more equitable because of the fact that individuals choose optional lifestyle choices and therefore put themselves in higher risk categories.¹⁰⁵ For example, a smoker pays a higher premium than a non-smoker for life insurance.¹⁰⁶ It has been observed that insurance premiums are based “*on a principle of equity, not equality*,” meaning that an insured’s predicted risk will dictate the premium charged.¹⁰⁷ Pursuant to this principle, a premium will be lower if the estimated risk is lower.

From a business perspective, risk classification facilitates the maximisation of profits by offering different rates based upon risk. Insurers therefore engage in competition by offering lower insurance rates to what are deemed to be lower risk

¹⁰¹ For further discussion of the concept of risk classification, see Kenneth Abraham, ‘Efficiency and Fairness in Insurance Risk Classification’ (1985) 71 Virginia Law Review 403

¹⁰² Robert F. Rich and Julian Ziegler, ‘Genetic Discrimination in Health Insurance – Comprehensive Legal Solutions for a (Not so) Special Problem?’ (2005) 2 Indiana Health Law Review 5, 11-12. See also Karen A. Clifford and Russell P. Incalano, ‘AIDS and Insurance: The Rationale for AIDS- Related Testing’ (1987) 100 Harvard Law Review 1806, 1810

¹⁰³ Roberta M. Meyer, ‘Justification for Permitting Life Insurers to Continue to Underwrite on the Basis of Genetic Information and Genetic Test Results’ (1993) 27 Suffolk University Law Review 1271, 1287

¹⁰⁴ Robert F. Rich and Julian Ziegler, ‘Genetic Discrimination in Health Insurance – Comprehensive Legal Solutions for a (Not so) Special Problem?’ (2005) 2 Indiana Health Law Review 5, 12

¹⁰⁵ Eric M. Holmes, ‘Solving the Insurance/ Genetic Fair/ Unfair Discrimination Dilemma in Light of the Human Genome Project’ (1996- 1997) 85 Kentucky Law Journal 3 503, 533

¹⁰⁶ Ibid

¹⁰⁷ Ibid

customers.¹⁰⁸ Indeed, it has been pointed out that, “*in order to stay in business,*” it is necessary for insurance companies to discriminate between individuals by means of risk classification.¹⁰⁹ In this regard, it has been observed that promoting efficiency by means of risk classification can often mean sacrificing other values, for example, equality and access to health care.¹¹⁰ Ethical issues arise here in circumstances where high risk individuals are unable to afford high premiums, and are therefore unable to access insurance. Having established that insurance companies generally engage in differentiation practices as part of fundamental principles and with a view to maintaining the economic viability of the industry, this provokes consideration of the impact which use of genetic information might have in this context.

6.4 Impact of genetic information on the insurance industry

Individuals who know that they have a heightened risk or predisposition to a certain condition may have a increased incentive to increase their insurance coverage accordingly. This may result in an imbalance in the information provided. Those in favour of use of genetic test results in the insurance industry argue that genetic information has to be accessible to insurers “*as a matter of equity,*”¹¹¹ to facilitate accurate risk classification, maintain the economic viability of the industry, and in accordance with the principle of utmost good faith (as discussed above). Pursuant to the principle of uberrimae fides, the failure to disclose relevant details of one’s family history of illness or the results of genetic tests previously taken may be deemed to be in breach of fundamental insurance principles. In this regard, the following section will highlight the concept of adverse selection.

6.5 Adverse selection

Adverse selection describes “*any process that results in an undesirably high proportion of high- risk insureds purchasing insurance coverage from a particular insurance company.*”¹¹² The process can take place in circumstances where an

¹⁰⁸ Herman T. Bailey *et al*, ‘The Regulatory Challenge to Life Insurance Classification’ (1976) 25 Drake Law Review 779, 782. See also Kenneth Abraham, *Distributing Risk: Insurance, Legal Theory and Public Policy* (Yale University Press 1986)

¹⁰⁹ Richard F. Rich and Julian Ziegler, ‘Genetic Discrimination in Health Insurance – Comprehensive Legal Solutions for a (Not so) Special Problem?’ (2005) 5 Indiana Health Law Review 5, 12. See also Jennifer S. Geeter ‘Coding for Change: The Power of the Human Genome to Transform the American Health Insurance System’ (2002) 28 American Journal of Law and Medicine 1

¹¹⁰ Kenneth Abraham, ‘Efficiency and Fairness in Insurance Risk Classification’ (1985) 71 Virginia Law Review 403, 404

¹¹¹ Robert J. Pokorski, ‘Use of Information by Private Insurers’ in Thomas F. Murphy and Marc A. Lappe (eds) *Justice and the Human Genome Project* (California University Press 1994) 91

¹¹² Roberta M. Berry, ‘The Human Genome Project and the End of Insurance’ (1996) 7 University of Florida Journal of Law and Public Policy 2 205, 217

individual applicant does not disclose relevant details of their risk factors.¹¹³ It is the purchase of insurance by applicants who represent higher risks than the insurers are aware of.¹¹⁴ Adverse selection may be cited as a reason in favour of use of genetic information in the insurance industry.¹¹⁵ Upon uncovering details of one's genetic make- up, customers may decide to take out additional insurance coverage to cover their increased risk. In these cases, there is a potential imbalance in information resulting in inaccurate actuarial calculations, with the consequence that the premiums collected will not be sufficient to cover the claims. This practice could eventually lead to the collapse of the insurance industry.

The question then arises as to whether adverse selection is in fact a reality in the insurance industry or whether it is merely a potential future risk. Studies have shown that adverse selection is in fact a reality. One study in particular found that women who had tested positive for carrying the breast cancer gene sought increased life insurance.¹¹⁶ This indicates that individuals are interested in taking advantage of their knowledge or increased mortality risk, that insurance companies are not aware of these risks, and therefore adverse selection is a real consideration.¹¹⁷ It is therefore an important consideration in this debate, from the perspective of insurers.

Insurance companies deal with adverse selection in a number of ways. It is noted that adverse selection could be cross- subsidised by large insurance pools. Cross- subsidisation is the process whereby insurance companies charge higher prices to one group of consumers with a view to subsidising lower prices for another group.¹¹⁸ Insurance companies address potential adverse selection and cross- subsidisation by engaging in risk classification in the underwriting process.¹¹⁹ To carry out this process, the insurance company will ask individuals questions about their health status, including requesting medical records, as well as family history. Knowledge of genetic information by insureds may further increase the imbalance of information and the potential of adverse selection. This raises the question whether genetic information should be available to insurers in the underwriting process, and under what terms. The next section will further explore the implications of using genetic information in the insurance industry.

¹¹³ Ibid. See R. Strohmer and A. Wambach, 'Adverse selection and categorical discrimination in the health insurance markets: the effects of genetic tests' (2000) 19 *Journal of Health Economics* 197-218

¹¹⁴ Ibid

¹¹⁵ Ibid

¹¹⁶ Cathleen D. Zick *et al*, 'Genetic testing, adverse selection, and the demand for life insurance' (2000) 93 *American Journal of Medical Genetics* 1 29- 39. See also Katrina Armstrong *et al*, 'Life insurance and breast cancer risk assessment: adverse selection, genetic testing decisions, and discrimination' (2003) 120(A) *American Journal of Medical Genetics* 3 359- 364

¹¹⁷ Ibid

¹¹⁸ See generally Robert Puelz, 'Evidence on Adverse Selection: Equilibrium Signaling and Cross-Subsidization in the Insurance Market' (1994) 102 *Journal of Political Economy* 2 236

¹¹⁹ Ibid.

6.6 Arguments in favour of using genetic information

Insurers may argue that genetic information will facilitate improved underwriting by engaging in more accurate risk classification, reduce adverse selection, and result in a more efficient system of insurance.¹²⁰

Lenox outlines the potential adverse effects of prohibiting use of genetic information. He argues that prohibiting genetic information in the insurance industry could result in significant costs to insurers and the insured.¹²¹ Prohibiting the use of genetic information in the underwriting process could result in a prohibition in using almost all health information for the purpose of underwriting. Broad definitions of genetic tests and genetic information, may include all medical tests, including such traditionally used underwriting tools as testing for high cholesterol, cancer and diabetes. Such a prohibition may prevent insurers from requesting family history information.¹²² This may result in insurance companies allocating risk to other groups, resulting in additional costs to insurers and insureds, affecting the economic viability of the industry.¹²³

It is also argued that limiting the use of genetic information in underwriting could increase rather than reduce unfairness in the underwriting process.¹²⁴ If an insurer were obliged to disregard an applicant's increased level of risk as revealed by genetic information or test results, this could result in unfair discrimination against both the healthy and unhealthy individuals who may carry genes that have not yet been detected. Meyer opines that such a requirement would result in "*preferential treatment of individuals with genetic disease*."¹²⁵ It has been noted that insurance rates for healthy insureds would increase to cover losses generated from inaccurately low rates charged to individuals with genetic conditions. It has been argued that allowing insurers to use genetic information in underwriting also increases rather than reduces the availability of insurance coverage.¹²⁶ In this regard, it may also raise the question as to "*who deserves our solidarity?*"¹²⁷

¹²⁰ Alissa Johnson, 'Genetics and Health Insurance', *Genetics Brief*, June 2002, at www.ncsl.org/programs/health/genetics/geneticshealthins.pdf (accessed 25 April 2012)

¹²¹ Bryce A. Lenox, 'Genetic Discrimination in Insurance and Employment: Spoiled Fruits of the Human Genome Project' (1998) 23 *University of Dayton Law Review* 1 189, 198

¹²² *Ibid*

¹²³ Roberta M. Meyer, 'Justification for Permitting Life Insurers to Continue to Underwrite on the Basis of Genetic Information and Genetic Test Results' (1993) 27 *Suffolk University Law Review* 1271

¹²⁴ *Ibid* at 1287

¹²⁵ *Ibid*

¹²⁶ *Ibid* at 1281

¹²⁷ Ine Van Hoyweghen, 'Genetics and Insurance: new technologies, new policies, new responsibilities' in G de Vries, K Horstman (eds) *Genetics from Laboratory to Society: Societal Learning as an Alternative to Regulation* (Palgrave Macmillan 2008) 118- 137

It is noted that the more insurers are able to accurately classify risks, the more accurately they can calculate insurance premiums to everyone's advantage.¹²⁸ Therefore the argument could be made that genetic tests will increase the availability of insurance rather than perpetuating unfair discrimination. However, the other side of this argument would argue that it is more equitable to differentiate on the grounds of optional lifestyle risks, than on the grounds of immutable characteristics such as genes. It is nevertheless worthwhile to make the observation as to which risks merit protection, and how the protection of one set of risk factors may negatively impact upon another set of factors.

Rothschild and Stiglitz also argue that, in the case of insurance, additional information is to be welcomed.¹²⁹ In circumstances where less reliable information is available to insurers, the practice of cross- subsidisation will continue to take place, potentially with adverse implications for the economic viability of the industry. Riba advises that this pattern could result in a disruption in the market, as the pricing system used by insurance companies would become ineffective.¹³⁰ In such scenarios, low- risk individuals may begin to view insurance as optional and may stop purchasing insurance. As a result of this adverse selection, insurers may assume that only high- risk people remained in their pool and would price policies accordingly, effectively pushing individuals out of the market.¹³¹ However, Riba neglects to consider the limited accuracy and predictive value of most genetic tests. This is a key point in the context of insurance. Vague information, indicating only an higher probability of developing a certain disease or disability, will not operate to increase the accuracy of an insurance company's risk prediction. Indeed such information might also be misinterpreted by insurers, with adverse effects on some insurance applicants.

The argument is sometimes offered that prohibitive legislation might stand in the way of favourable use of genetic information. It is noted that genetic testing may improve the insurability of people who have difficulty obtaining insurance because of family history – for example, family history of Huntington's disease.¹³² This provokes the question – in certain circumstances, should insurance companies have access to negative genetic test results and indeed should it be permissible to volunteer negative genetic test results. It seems logical to assume that in the same way that a positive test result for a genetic condition may hinder someone's insurance application, a negative test result may very well be looked upon favourably by an insurance company. There are therefore compelling arguments in favour of use of genetic information by insurers, primarily in

¹²⁸ See generally Naomi Obinata, 'Genetic Screening and Insurance: Too Valuable a Tool to be Banned from the System' (1992) 8 Santa Clara Computer and High Technology Law Journal 145

¹²⁹ Michael Rothschild and Joseph Stiglitz, 'Equilibrium in Competitive Insurance Markets: An Essay on the Economics of Imperfect Information' (1976) The Quarterly Journal of Economics 629, 629

¹³⁰ Shannyn C. Riba 'The Use of Genetic Information in Health Insurance: Who will be Helped, who will be harmed and the possible long- term effects' (2007) 16 Review of Law and Social Justice, 2 470, 480

¹³¹ Ibid

¹³² Ibid

consideration of the economic viability of insurance companies and fundamental principles of insurance.

6.7 Adverse implications of using genetic information

The primary reason offered opposing an insurer's use of genetic information is the potential misuse of such information, leading to discrimination and breach of privacy. First of all, unfair discrimination may take place. Using genetic information in underwriting may potentially result in further classification of individuals, and perhaps act as a barrier to accessing insurance. Those who test positive for the presence of a certain gene, or those who have a family history of certain conditions may be classified as "*high-risk*" candidates.¹³³ As a result, there is a possibility that these individuals may either be denied insurance cover, or charged inordinately high premiums thereby potentially pushing them out of the insurance pool. By allowing the free use of genetic information in insurance, there is a possibility that insurers will take advantage of this information, leading to misuse.¹³⁴ Further, the argument can be made that it is unfair to discriminate on the basis of immutable characteristics such as genes, over which the individual has no control, and did not choose.

In addition, concerns also arise as regards the right to privacy and the right to respect of medical confidentiality.¹³⁵ Genetic information contains personal details of an individual's health status. Unauthorised access to and use of genetic information may violate an individual's privacy with adverse implications for psychological, emotional and perhaps physical wellbeing. As emphasised, one of the unique features of genetic information is that it reveals sensitive, personal information about not just an individual, but that individual's family. There are therefore additional privacy and confidentiality concerns that arise, when considering the use of genetic information.

There are further societal implications. Pushing individuals out of insurance pools by refusing insurance or charging high premiums may create barriers to not only to insurance, but also may create barriers to other social goods and services. This may result in the isolation of certain vulnerable individuals from society (particularly persons with putative disabilities and elderly persons). This may further lead to the exclusion of such vulnerable persons from health services, property ownership and other social goods and services. Ultimately this may lead to the creation of a genetic underclass. There are compelling reasons to consider the appropriate regulation of the area.

¹³³ See generally Lawrence Gostin, 'Genetic Discrimination: The Use of Genetically Based Diagnostic and Prognostic Tests by Employers and Insurers' (1991) 17 *American Journal of Law and Medicine* 109

¹³⁴ Eric M. Holmes, 'Solving the Insurance/ Genetic Fair/ Unfair Discrimination Dilemma in Light of the Human Genome Project' (1996- 1997) 85 *Kentucky Law Journal* 3 503, 507

¹³⁵ For further discussion see Mark Taylor, *Genetic Data and the Law: A Critical Perspective on Privacy Protection* (Cambridge University Press 2012)

6.8 The social purpose of insurance

As highlighted, in mutuality based insurance systems, insurers differentiate between individuals on the basis of risk assessment, often resulting in the refusal to insure what are perceived as high risks (or the imposition of high premiums). As a result, access to health care for persons with high risks may be effectively blocked and access to other insurance products such as life insurance may also be inhibited. This is undesirable from the view that insurance is important and indeed necessary for all individuals.

In particular, it is generally recognised that health care is such an important social good that access to it should be guaranteed to everyone.¹³⁶ Murray refers to the importance of maintaining the social purpose of insurance, and also observes that genetic differences do not constitute appropriate reasons for treating people differently with respect to accessing health care.¹³⁷ It follows that treating individuals differently and engaging in the process of risk classification, acts as a barrier to universal access to health care. It has been pointed out that the concept of actuarial fairness is morally questionable, and (apart from economic arguments), it is difficult to reconcile the wish of insurers to use genetic information with the need of individuals to access health care and insurance.¹³⁸

Life insurance, although generally not considered to be as essential a social good as health insurance equally merits consideration as an important element of an individual's private life.¹³⁹ Life insurance can provide peace of mind and security for individuals and their dependents. In this respect, it has been observed that life insurance is perceived as "*serving a need rather than being a mere commodity*"¹⁴⁰ and ought to be accessible, without barriers. It has also been described as a "*non- primary social good*" which should be accessible to as many people as possible.¹⁴¹

In addition, in jurisdictions where there may be a requirement to purchase life insurance as a prerequisite to obtaining a mortgage, life insurance is further viewed as essential in securing fundamental human rights and providing a

¹³⁶ Trudo Lemmens, 'Selective Justice, Genetic Discrimination, and Insurance: Should we Single Out our Genes in Our Laws?' (2000) 45 McGill Law Journal 347, 388-389

¹³⁷ Thomas Murray, 'Genetics and the Moral Mission of Health Insurance', 22 Hastings Center Report 6, 00930334, (November 1992)

¹³⁸ Ibid

¹³⁹ Janneke H. Gerards and Heleen L. Janssen, 'Regulation of Genetic and Other Health Information in a Comparative Perspective' (2006) 13 European Journal of Health Law 339, 359

¹⁴⁰ Thomas Murray, 'Genetics and the Moral Mission of Health Insurance' (November 1992) 22 Hastings Center Report 6, 00930334

¹⁴¹ Per Sandberg, 'Genetic Information and Life Insurance: A Proposal for an Ethical European Policy' (1995) 40 Society Science & Medicine 11 1549, 1554

gateway to property ownership.¹⁴² In circumstances where the purchase of property, (or other social goods) is dependent upon having a certain type of insurance, there is an additional value placed on the importance of insurance. Insurance coverage may therefore be seen as important for one's full participation in society.

Therefore, it is arguable that access to genetic information by insurers may lead to undue interference with an individual's right to access health care and insurance. The importance of recognising insurance as a key social good therefore adds merit to the hypothesis that the area of insurance ought be regulated to prevent misuse of genetic information and to ensure that individuals are not pushed out of insurance pools. This is a particularly strong argument in the context of health insurance. Consideration of the social purpose of insurance results in a conflict of rights between the moral argument that everyone should have access to insurance, and the economic argument that insurers treat individuals differently. It is acknowledged that the economic rationale underpinning the insurance industry is a strong consideration in this debate, particularly in the life insurance industry. The social purpose of insurance is nevertheless a relevant factor to consider when seeking to strike a balance between the competing interests.

6.9 Recent developments in the insurance industry

As mentioned, discussion of genetic discrimination in the insurance industry gives rise to a tension between various competing rights. It is submitted that insurance law potentially comes into conflict with the principles of non-discrimination. This is particularly evident in a recent European Court of Justice (ECJ) case concerning gender as a risk factor.¹⁴³ In the *Test- Achats* case, the Belgian consumer group successfully challenged Article 5 (2) of the Gender Equality Directive,¹⁴⁴ arguing that it is in conflict with the EU principle of equal treatment of men and women. In its reasoning the ECJ held that the derogation in Article 5 (2) of the Directive, which allows insurers to vary insurance premiums or benefits according to gender if based on relevant statistical data, is contrary to the fundamental principle of equal treatment between the sexes.

¹⁴² See generally Mark A. Rothstein and C.A. Hornung, 'Public Attitudes' in Mark A. Rothstein (ed) *Genetics and Life Insurance. Medical Underwriting and Social Policy* (The MIT Press 2004) 23. Trudo Lemmens, 'Selective Justice, Genetic Discrimination, and Insurance: Should we Single Out our Genes in Our Laws?' (2000) 45 McGill Law Journal 347, 391

¹⁴³ Case C- 236/09 *Association Belge des Consommateurs Test- Achats ASBL v Conseil des ministres* [2011] (1 March 2011) ECR I- 00773. See Eugenia Caracciolo di Torella, 'Gender Equality after Test Achats' 13 ERA Forum (Springer 2012) 59. See also Vadim Mantrov, 'Gender Factor in the Insurance Law: Recent Development of the ECJ in Context with the U.S. Approach' (accessed 26 February 2012) Available at: <http://ssrn.com/abstract=2028992>

¹⁴⁴ Council Directive 2004/113/EC of 13 December 2004 implementing the principle of equal treatment between men and women in the access to and supply of goods and services [2004] OJ L 373/37 ("Gender Equal Treatment Directive")

This case will have a significant effect on insurance industry practices and risk assessment. Since 21 December 2012, insurers in the EU may not use gender as a risk factor in setting insurance rates. The judgment is illustrative of the ECJ's intention to expand the reach of equality principles, and focus on the importance of recognising fundamental human rights. This also provokes consideration of whether it is foreseeable that discrimination on other grounds such as age, disability or genetic information could also be deemed to be prohibited conduct by insurance companies in the future. On the basis of the rationale advanced in the *Test- Achats* case, this might not be outside the realms of possibility. Indeed it certainly forces us to recognise the prime position taken by fundamental human rights of equality and non- discrimination in the EU. This case, and its implications will be discussed in greater detail in chapter 9.

6.10 Reaching a balance

The use of genetic information must be considered from both the individual's and the insurer's perspective, balancing the economic based right to know of the insurance company against the individual's right to privacy, right not to be discriminated against and to have access to insurance. It is submitted that insurance law comes into conflict with the principles of non- discrimination and equality, as illustrated in the recent ECJ case concerning gender as a risk factor.

In looking towards a balance, it has been observed that in order for the private insurance industry to maintain stability, a certain degree of risk classification is needed and must be accepted by all stakeholders.¹⁴⁵ This submission is most relevant as regards life insurance. It is necessary therefore to consider a balance between the competing interests of both insurers and insureds – to ensure fairness of contract and to uphold the fundamental principles of insurance law. It is important that the social purpose of insurance is also considered – to ensure that those who need the benefits which insurance affords are not pushed out of the insurance pools.

This need for a balance raises the question of the extent of an insurer's right to access genetic information. Firstly, an insurer might request that an individual undergo a particular genetic test. Secondly, an insurer might request disclosure of genetic test results previously taken by the individual. Thirdly, an insurer might request disclosure of an individual's family history information.

As regards life insurance, it is submitted that an insurer should not be permitted to demand that an individual undergo a genetic test in any circumstances. Such a request would conflict with an individual's fundamental right to autonomy and self- determination, as well as adversely affect the right not to know. It would also reveal information that was previously unknown to both the insurer and the

¹⁴⁵ Yann Joly *et al*, 'Genetic discrimination in private insurance: global perspectives' (2010) 29 New Genetics and Society 4 351, 365

insured, therefore it would not impact upon the duty of utmost good faith. In relation to accessing results of genetic tests previously taken or family medical history, it is acknowledged that a balance needs to be reached. It is arguable that insurers should be permitted to request such information, but under strict conditions. With a view to facilitating the competing rights, insurers should only be permitted to request such information, in the context of life insurance, where the insurance cover sought is above a certain amount, and where the accuracy of the information is actuarially and statistically valid. As regards health insurance, it is submitted that an insurer should not be permitted to demand that an individual undergo a genetic test, request the results of genetic tests previously taken, or request family medical history in any circumstances. This submission is particularly in consideration of the importance of health insurance as a vital social good.

Importantly, any consideration of third party use of genetic information must therefore take into account the limited accuracy and predictive value of most genetic tests, and acknowledge the distinction between monogenic and multifactorial genetic conditions. Indeed, this may point towards the need for an education campaign to inform individuals and third parties as to the reality of advancing genetic science and technology, including its potential benefits as well as its limitations.

In addition, it is noted that insurers must recognise how their practices create public apprehension, suspicion and controversy.¹⁴⁶ It is submitted that there is a need for greater transparency in the insurance industry, particularly in relation to risk assessment and underwriting practices. There is a general distrust and suspicion surrounding the insurance industry, and this will undoubtedly be exacerbated in this new age of advancing genetic science. In order for individuals to trust insurance companies, it is important that there is public engagement with the insurance industry, and that insurance practices are open and transparent.

6.11 Evaluation of third party interests

Third party interest in genetic information stems primarily from an economic interest, and the desire to use such information to fulfill financial objectives. Unfettered access to and use of genetic information by third parties can result in discrimination and breach of privacy, with further ethical concerns for the individual, family members and society. It is therefore important to highlight the tension that arises between the competing rights. The primary focus of this thesis is on unauthorised use of genetic information by third parties such as employers and insurers, and the ethical and legal dilemmas this gives rise to. This debate therefore needs to be informed of the conflict of rights arising, with a view to ascertaining the appropriate means of regulation. The following part of this

¹⁴⁶ Ibid

chapter will look at the societal interest in this area, and how it can be affected by misuse of genetic technologies.

7. Societal interest in genetics and advancing technology

This section will highlight the societal interest in genetic science. This interest primarily concerns the desire to enhance public health as well as the need to ensure the appropriate application of science and technology.

7.1 Public health interest

Genetic science and advancing genetic technology has become a powerful tool to understand, and potentially improve health and disease. Society therefore has an interest in genetic advances primarily from a public health perspective, and in ensuring enhanced health care for all individuals. A healthier population and an improved public health care system have economic benefits for society, primarily by promoting more effective and efficient diagnosis and treatment of disease. The general benefits brought about by advances in genetics such as a greater understanding of disease and its treatment, as well as the promise of personalised medicine, stand to benefit the entire population. Society therefore has a clear interest in the overall positive benefits of genetic technology.

In line with this interest, it is acknowledged that scientists and researchers require the use of genetic information, and rely upon individuals engaging in genetic testing and clinical trials. Without access to such information and in the absence of participation in clinical trials, it may prove challenging to identify connections between genes and disease. This universal benefit reinforces the societal interest in allowing genetic science to advance and develop. It is observed that competing rights and interests in genetic information may operate to impact upon the societal interest in genetic information. It is noted that the fear of the misuse of one's genetic information, results in some individuals being reluctant to take advantage of advancing genetic technology.¹⁴⁷

This reluctance to engage in genetic testing may also have the effect of stifling science and technology. Scientists may therefore experience difficulty in collecting research data as people become unwilling to submit to testing and engage in clinical trials. This may have further implications for public health, as potentially beneficial genetic discoveries and technological innovations become hampered. These issues raise serious public policy concerns that must be considered as part of this debate. It also highlights the need to ensure that public confidence in the area is addressed.

¹⁴⁷ Louise Slaughter, 'The Genetic Information NonDiscrimination Act: Why your personal genetics are still vulnerable to discrimination' (2008) 88 The Surgical Clinics of North America 4 723, 737

Society also has an interest in ensuring the appropriate and lawful use of genetic science and technology, as well as ensuring that such technology is not abused. This section will focus on the need to ensure appropriate application of genetic science.

7.2 Ethnic and racial discrimination

This section will highlight how genetic discrimination may perpetuate further discrimination against already marginalised and historically victimised groups, such as racial and ethnic minorities.¹⁴⁸ Genetic conditions are sometimes more prevalent in certain racial and ethnic groups, or in specific communities.¹⁴⁹ For example, the sickle-cell trait generally affects Africans and African-Americans at a higher rate than other races.¹⁵⁰ The BRCA 1 and BRCA 2 genes are more common among Ashkenazi Jews.¹⁵¹ Similarly, Tay-Sachs disease is more prevalent among Ashkenazi Jews than any other race or ethnicity.¹⁵² As advancing science is identifying the genetic basis of a wide range of conditions, the prevalence of genetic conditions amongst certain groups is becoming more apparent. Genetic discrimination might also perpetuate further discrimination between the sexes, for example, in the employment context. It is observed that a high rate of a particular genetic predisposition or condition in certain populations could be used to stigmatise the entire group. Third parties might recognise the genetic link between groups, and discriminate based on certain perceptions they might have, or based on an existing stigma attached to that group.¹⁵³

¹⁴⁸ Lawrence Gostin, 'Genetic Discrimination: The Use of Genetically Based Diagnostic and Prognostic Tests by Employers and Insurers' (1991) 17 *American Journal of Law and Medicine* 109, 111. See generally Iulia V. Motoc, 'The International Law of Genetic Discrimination: The Power of Never Again' in Therese Murphy (ed) *New Technologies and Human Rights* (Oxford University Press 2009)

¹⁴⁹ Anne Mainsbridge, 'Employers and Genetic Information: A New Frontier in Discrimination' (2002) 2 *Macquarie Law Journal* 61, 71. There has recently been controversy in relation to screening for sickle cell anaemia in the NCAA – for further discussion see, Lanetta B. Jordan *et al*, 'Screening U.S. College Athletes for their Sickle Cell Disease Carrier Status' (2011) 41 *American Journal of Preventative Medicine* 6 406. See also, Francis S. Collins, 'What we do know and what we don't know about 'race', 'ethnicity', genetics and health at the dawn of the genome era' (1994) 36 *Nature Genetics Supplement* 11

¹⁵⁰ See generally Trudo Lemmens, 'Selective Justice, Genetic Discrimination, and Insurance: Should we Single Out our Genes in Our Laws?' (2000) 45 *McGill Law Journal* 347, 372- 373

¹⁵¹ See Shirley V. Hodgson *et al*, 'Risk Factors for Detecting Germline BRCA1 and BRCA2 Founder Mutations in Ashkenazi Jewish Women with Breast or Ovarian Cancer' (1999) 36 *Journal of Medical Genetics* 369

¹⁵² See generally Rachel Myerowitz and F. Christine Costigan, 'The Major Defect in Ashkenazi Jews with Tay-Sachs Disease Is an Insertion in the Gene for the a-Chain of b-Hexosaminidase' (1988) 263 *The Journal of Biological Chemistry* 35 18587. See also John J. Mitchell *et al*, 'Twenty- Year Outcome Analysis of Genetic Screening Programs for Tay- Sachs and b- Thalassemia Disease Carriers in High Schools' (1996) 59 *American Journal of Human Genetics* 793, Barbara L. Triggs- Raine *et al*, 'Screening for Carriers of Tay Sachs Disease among Ashkenazi Jews – A Comparison of DNA Based and Enzyme Based Tests' (1990) 323 *The New England Journal of Medicine* 6

¹⁵³ Henry T. Greely, 'Legal, Ethical, and Social Issues in Human Genome Research' (1998) 27 *Annual Review of Anthropology* 473, 483

Indeed we can look to history to illustrate the potential for abuse of science and the vulnerability of certain groups. It has been observed that historically, similar abuses of science have given rise to undesirable social practices with the objective of weeding out individuals perceived to have certain defects. For example, historically the Ashkenazi Jews were stigmatised and killed in Nazi Germany (discussed below). Potentially, screening for breast cancer and Tay-Sachs disease may negatively target Ashkenazi Jews and a genetic test result may result in further marginalisation of this historically stigmatised minority. As science advances, more sophisticated links between genetic diseases and populations are inevitably going to be discovered. This can result in increased discrimination, social exclusion and marginalisation, in employment, insurance and in society.

7.3 Evaluation of societal interests

The societal interest in genetic information is characterised by an overall interest in the application of genetic science and technology for the benefit of all. It is observed that society also has an interest in the appropriate application of this science, acknowledging the potential for misuse of genetic information. It is further acknowledged that other competing interests in genetic information may impact upon the overall societal interest. It is observed that an individual exercising their rights may have the result of stifling science and technology, with negative societal consequences. This indicates the need to ensure that public confidence in this area is enhanced. The societal interest also provokes further public policy issues, which will be discussed in the following section.

8. Public policy concerns

In addition to examining the ethical concerns arising in this area, it is also necessary to highlight broader public policy concerns which shape this debate, and allow us to anticipate the potential for abuse of advancing genetic technologies. This section will illustrate how advances in genetic science and our understanding of disease and disability can impact upon notions of personhood and individual identity. The potential for an over-reliance on genetic science can lead to the expression of theories of genetic determinism, with possible eugenic consequences.

8.1 Personhood considerations

The concept of personhood has been explored extensively in literature and philosophy. It is submitted that the notion of personhood can be influenced by advances in genetics, and it is interesting to examine the relationship between genetic information and personal identity. First, it is necessary to define 'personhood.' Personhood is differentiated from the notion of being human, which

means to belong to a species.¹⁵⁴ Personhood represents one's recognition as a person – and “*therefore as a ‘subject’ of the law and the political order – as a beneficiary of the system of justice.*”¹⁵⁵ Concepts of personhood have evolved over time, particularly with shifts in societal and cultural norms. For example, Quinn has pointed out that personhood was limited by the Romans to males,¹⁵⁶ and Blackstone opined that upon marriage, women suffered a civil death in the sense that her personhood was merged with her husband's.¹⁵⁷

In this new genetic age, with scientists making rapid discoveries about genes, it is likely, that concepts of personhood will further develop and evolve. Genetic information and the human genome relate to the biological component of personhood, and how this impacts on our overall conception of what it means to be a person.¹⁵⁸ An individual's genetic information determines a range of characteristics and traits from height, skin colour, eye colour, gender, behaviour and personality. With scientific research revealing core elements of a human's unique make up and other personal traits, this will inevitably have an impact on conceptions of self.¹⁵⁹

The impact of genetic information on personal identity accordingly provokes consideration of the concepts of genetic determinism and genetic reductionism.¹⁶⁰ With the focus on the importance of genes on the expression of disease and disability, there is an increasing potential for promotion of the “*geneticized self*”, in other words reducing one's identity to a genetic basis.¹⁶¹ These notions of the “*geneticized self*” raise ethical concerns, particularly in relation to notions of humanity. With advancing genetic science comes an increasing opportunity to enhance oneself, with implications for human dignity and uniqueness. It is observed that such conceptions of self do not acknowledge the whole person. Within the genetic conception of self, “*the person then becomes no more than their ‘bad’ genes,*”¹⁶² potentially leading to discrimination and stigmatisation.

Certain theories of genetic determinism suggest that our genetic profile is “*the dominant causal arbiter of all that we think, feel, and choose to do.*”¹⁶³ Taken to the extreme, genetic determinism implies that a person does not possess the

¹⁵⁴ Gerard Quinn, ‘Rethinking Personhood: New Directions in Legal Capacity Law and Policy’ (University of British Columbia, Vancouver, Canada) (29 April 2011)

¹⁵⁵ Ibid

¹⁵⁶ Ibid

¹⁵⁷ William Blackstone, *Commentaries on the Laws of England* (15th edn, London 1809) 441- 442

¹⁵⁸ Hugh Miller, ‘DNA blueprints, personhood and genetic privacy’ (1998) 8 Health Matrix 179, 187

¹⁵⁹ Ibid

¹⁶⁰ For further discussion of the theory of genetic determinism, see George P. Smith II and Thaddeus J. Burns, ‘Genetic Determinism or Genetic Discrimination?’ (1994- 1995) 11 Journal of Contemporary Health Law and Policy 23

¹⁶¹ Jennifer Fitzgerald, ‘Geneticizing Disability: The Human Genome Project and the Commodification of Self’ (1998- 1999) 14 Issues in Law and Medicine 2 147, 151

¹⁶² Ibid at 151

¹⁶³ Hugh Miller, ‘DNA blueprints, personhood and genetic privacy’ (1998) 8 Health Matrix 179, 207

freedom to think, or choose. This conception is contrary to the interpretation of a person as an autonomous being who can freely choose between various courses of action.¹⁶⁴ Strict application of the theory of genetic determinism denies that individuals have the power of free choice. In response to this, there is a compelling practical reason for maintaining the concept of free will and rejecting genetic determinism. Such a theory would “*wreak havoc on our ordinary conceptions of ethically responsible agency and moral desert.*”¹⁶⁵

In reality, a well-informed view of genetics does not support the theory of genetic determinism. The key to the nature of personal identity is to be found in the myriad of personal circumstances and facts that make up each person’s life. For example, a person’s environment, occupation, and lifestyle choices contribute to one’s sense of self. In addition, only a few genetic conditions are solely determined by a certain gene or genes.¹⁶⁶ Accordingly, “*genes are only one more influence in the contingent histories of our lives.*”¹⁶⁷ From a human rights perspective, the concept of the “*geneticized self*” threatens an individual’s basic human rights particularly since they are then “*defined out of humanity and potentially outside the boundaries of moral responsibility.*”¹⁶⁸ There is also a danger of an individual’s unique identity becoming lost and their lives becoming devalued. These notions reflect the core of the concept of personhood and the concept of recognising the legal capacity of all individuals.

From a public policy perspective, this reality of genetic science points to a need to educate physicians, lawyers, policy makers, as well as the general public, to garner an understanding of the realities of science, its possibilities, but also its limitations. In the absence of such an understanding, and appropriate education, and in consideration of the speed at which science is advancing, there is a real potential for the misinterpretation of science, and the development of negative attitudes toward personal identity. It is submitted that theories of genetic determinism have the potential to produce eugenic effects, and can ultimately lead to the creation of a genetic underclass.

8.2 A potential genetic underclass

Society may eventually view individuals on the basis of their genes, differentiating according to what are deemed to be good genes and bad genes, leading to discrimination and segregation. It has been observed that the motion picture, *GATTACA* illustrates the potential consequence of this genetic underclass: “*a world where a person’s only sense of identity comes from his or*

¹⁶⁴ Ibid at 205

¹⁶⁵ Ibid at 207

¹⁶⁶ Henry T. Greely, ‘Legal, Ethical, and Social Issues in Human Genome Research’ (1998) 27 Annual Review of Anthropology 473, 496

¹⁶⁷ Ibid

¹⁶⁸ Jennifer Fitzgerald, ‘Geneticizing Disability: The Human Genome Project and the Commodification of Self’ (1998- 1999) 14 Issues in Law and Medicine 2 147, 152

her genes,”¹⁶⁹ thereby reflecting a strong genetic determinist society. It illustrates how genetic discrimination operates to exacerbate racial and other segregation in society. Again, this may impact upon concepts of personhood and result in a society valued by genetic status, thereby disregarding a person’s inherent worth and recognition as a person.

We could potentially end up with such a ‘genetically cleansed’ society and economy whereby only those who are considered risk-free are admitted to the mainstream. Eugenic concerns arise in this context, and we can look to the past history of eugenics as an indicator of potential abuse in the future. It is noted that the “*relationship between the past and present uses of genetic knowledge illustrates the complex ways in which social, political, and ethical norms of the present interact with and shape our understanding of the past.*”¹⁷⁰ The following section will further explore the potential impact of genetics on concepts of eugenics.

8.3 The relationship between genetics and eugenics

It is observed that eugenics originated from the renewed scientific interest in biological inheritance that was prompted by the rediscovery of Mendel’s work at the end of the nineteenth century.¹⁷¹ Eugenics applied the principles of heredity established by Mendel with a view toward improving human beings.¹⁷² Eugenics has been described as the “*conscious selection of humans by encouraging the production of those with desired inherited characteristics and for restricting those with undesirable inheritable characteristics.*”¹⁷³ It can also be defined as “*the use of science applied to the qualitative and quantitative improvement of the human genome.*”¹⁷⁴ The subject was pioneered by Francis Galton, with reinforcement from Charles Darwin in the latter half of the 19th century.¹⁷⁵

Since the 19th century, there has been an acknowledgement of the benefits of using science and principles of inheritance to shape eugenic policies, with the result of creating a genetically cleansed society. Indeed this early recognition of the benefits of genetics resulted in an extreme application of this knowledge and

¹⁶⁹ See generally, David A. Kirby, ‘Extrapolating Race in GATTACA: Genetic Passing, Identity, and the Science of Race’ (2004) 23 Literature and Medicine 1, 184- 185

¹⁷⁰ Lene Koch, ‘The Meaning of Eugenics: Reflections on the Government of Genetic Knowledge in the Past and Present’ (2004) 17 Science in Context 3 315, 316. doi:10.1017/50269889704000158

¹⁷¹ Tamsen Wolff, ‘Mendel’s Theatre: Heredity, Eugenics and Early Twentieth Century American Drama’ (Palgrave Macmillan 2009). See also Karen H. Rothenberg, ‘From Eugenics to the “New” Genetics : “The Play’s the Thing”’ (2010) 79 Fordham Law Review 407, 409

¹⁷² Ibid at 95- 97

¹⁷³ Willi Rothley and Carlo Casini, ‘Ethical and legal problems of genetic engineering and human artificial insemination’, (EU Committee on Legal Affairs and Citizen’s Rights, 1990)

¹⁷⁴ David J. Galton and Claire J. Galton, ‘Francis Galton: and eugenics today’ (1998) 24 Journal of Medical Ethics 99, 99

¹⁷⁵ Ibid. See also Francis Galton, *Inquiries into human faculty and its development* (Dent and Sons 1907)

formed the basis of the eugenic policies in the US and Europe in the 20th century. Eugenics presents a radical case of how genetic knowledge and a misinterpretation of genetic science can be coupled with repressive public policy to deprive individuals of fundamental rights. The next section will examine these policies in greater detail.

8.4 A historical perspective of eugenic policies in Europe and the United States

The two eugenic frameworks that have been most frequently cited are those in Germany and the US, both of which began during the end of the 19th century. It has been observed that “*by remembering and understanding the past injustices and inhumanity of negative eugenics, further misuse of scientific information can be avoided.*”¹⁷⁶ It is pointed out that in both Germany and the US, the framework for the eugenics movement was the rediscovery of Mendel’s laws of genetics.¹⁷⁷ The aim of the eugenics movement was to promote and advance genetic superiority, and it endeavoured to achieve this primarily by sterilising the genetically undesirable.¹⁷⁸ Similarly, in the 20th century, the concept of biological determinism was utilised by eugenicists in the US and Germany to provide answers to many of the social defects at the time. These same eugenicists also believed that many medical conditions were based solely on our genes. It is noted that using this framework, these proponents then educated others about the perceived dangers and “*devised ways of controlling them, namely through involuntary sterilization and genocide.*”¹⁷⁹

In European history, the Nazi regime provides the most obvious framework for discussion of the dangers of eugenics. It has been noted that “*the Nazis famously misused genetic information in incredibly discriminatory, invidious, and ultimately genocidal ways.*”¹⁸⁰ The isolation and elimination of those deemed to be inferior and problematic in society was premised on genetics, leading to the objective of achieving the perfect race.¹⁸¹ During the early 20th century this movement focused increasingly on race, and when Hitler came to power in 1933, the regime was firmly in place, receiving serious endorsement from Hitler and the

¹⁷⁶ Kenneth L. Garver and Bettylee Garver, ‘The Human Genome project and Eugenic Concerns’ (1994) 54 American Journal of Human Genetics 148, 148

¹⁷⁷ Ibid at 150

¹⁷⁸ Iulia V. Motoc, ‘The International Law of Genetic Discrimination: The Power of Never Again’ in *New Technologies and Human Rights* Therese Murphy (ed) (Oxford University Press 2009)

¹⁷⁹ Kenneth L. Garver and Bettylee Garver, ‘The Human Genome project and Eugenic Concerns’ (1994) 54 American Journal of Human Genetics 148, 150

¹⁸⁰ George J. Annas, ‘Genetic Privacy: There Ought to be a Law’ (1999) 4 Texas Review of Law and Policy 9, 12

¹⁸¹ Peter Harper, ‘Huntington’s disease and the abuse of genetics’ (1992) 50 American Journal of Human Genetics 460- 464. See also Deirdre Madden, *Medicine, Ethics, and the Law* (Butterworths 2002) 347

Nazis.¹⁸² The first involuntary sterilisation law in Germany was passed in 1933, at first to sterilise the mentally and physically disabled.¹⁸³ This regime was later extended to sterilise the various 'undesirable' ethnic groups, such as the Jews and Poles, who were forced into concentration camps.¹⁸⁴ In addition, the T4 Euthanasia Program was initiated in 1939 in Nazi Germany. It had the objective of eliminating those with physical or mental disabilities, and other vulnerable members of society.¹⁸⁵

Similarly, eugenic policies were utilised in Scandinavian countries, with a view to improving the genetic desirability of the population. In 1934 and 1935 sterilisation legislation resulting in voluntary as well as compulsory sterilisation were passed in Denmark, Norway and Sweden.¹⁸⁶ The eugenics movement in these countries was focused on the interests of the community and to achieve this objective it controlled the rights of some individuals and advanced the rights of others.¹⁸⁷ It is observed that "*it took decades before reproductive control came within the reach of the individual citizen,*" and such laws were still in existence until the 1970s.¹⁸⁸ The policies in these Scandinavian countries were a form of eugenics based on principles of genetics, which had the result of infringing human rights, and were illustrative of the impact of eugenic policies in 20th century Europe.

In the US, the history of eugenics is also very much apparent and has prompted a sense of fear when it comes to science and genetics. Indeed the history of eugenics in the US, and the resulting apprehension surrounding genetic science has shaped the current regulatory regime addressing genetic information in the US, as well as informing the ethical debate.¹⁸⁹ Eugenic policies in the US also took place in the areas of immigration and family life. In the 1920s, state fairs sponsored "*Fitter Families*" contests which awarded prizes for "*Grade A individuals*" in the "*human stock*" category.¹⁹⁰

In the early 20th century, the eugenics movement was becoming accepted, both by Congress and in society. At this time many US states introduced sterilisation

¹⁸² See generally Paul Weindling, *The Sonderweg of German Eugenics: Nationalism and Scientific Internationalism* (Cambridge University Press 1989). See also Paul Weindling, 'The Survival of eugenics in 20th century Germany' (1993) 52 *American Journal of Human Genetics* 3 643.

¹⁸³ Kenneth L. Garver and Bettylee Garver, 'The Human Genome project and Eugenic Concerns' (1994) 54 *American Journal of Human Genetics* 148, 150

¹⁸⁴ *Ibid* at 150

¹⁸⁵ M. Michael Cohen, 'Overview of German, Nazi and Holocaust Medicine' (2010) 152A *American Journal of Medical Genetics* 3 687. See also Rael D. Strous, 'Hitler's Psychiatrists: Healers and Researchers Turned Executioners and Its Relevance Today' (2006) 14 *Harvard Review of Psychiatry* 1 30

¹⁸⁶ Lene Koch, 'The Meaning of Eugenics: Reflections on the Government of Genetic Knowledge in the Past and Present' (2004) 17 *Science in Context* 3 315, 320. doi:10.1017/50269889704000158

¹⁸⁷ *Ibid*

¹⁸⁸ *Ibid*

¹⁸⁹ GINA, s 2(2) and (3)

¹⁹⁰ Daniel J. Kevles, *In the Name of Eugenics: Genetics and the Uses of Human Heredity* (Harvard University Press 1985) 62

laws targeting what were perceived to be genetically defective populations, following early developments in genetic science. The policy argument promoting such sterilisation was that those individuals were costly for society.¹⁹¹ Indeed, thirty- two American states introduced compulsory eugenic sterilisation laws from 1907 to 1937, primarily with the objective of controlling the reproductive capacities of these genetically undesirable individuals.¹⁹² The US Supreme Court endorsed state- sponsored sterilisation in the name of eugenics in its landmark decision of *Buck v. Bell*, with Justice Oliver Wendell Holmes infamously stating “...three generations of imbeciles is enough...”¹⁹³

In the US, the eugenics movement had a racial associations as well. The more desirable population was perceived to be “*white Protestants of Northern European Stock*”, while the less desirable population consisted of “*blacks and Jewish and Catholic immigrants*.”¹⁹⁴ This was particularly evident in the immigration policies of the early 20th century.¹⁹⁵ Another more recent example of genetic discrimination, and one which further illustrates the racial concerns associated with genetics, began in the 1970s with the discovery of the gene associated with sickle cell anemia, which is found primarily found in African-American individuals. In the 1970s, federal and state governments sponsored screening policies for sickle cell anaemia that were directed towards African Americans, with a view to identifying those carrying the sickle cell anaemia gene. Even though these policies had a public health objective, with legitimate aims, it resulted in stigmatisation and discrimination on the grounds of race.¹⁹⁶ To alleviate this stigma and discrimination, Congress passed the National Sickle Cell Anemia Control Act in 1972.¹⁹⁷

These eugenic regimes are an example of how misuse of science and new technologies can therefore impact upon human rights, as well as producing a myriad of undesirable societal implications. These incidences of stigmatisation and eugenics highlight historical patterns of prejudice and discrimination in the US and Europe and serve as a stark example of the undesirable implications of and misapplication of genetic science. The eugenics regime indicates how

¹⁹¹ Lori B. Andrews, ‘Past as Prologue: Sobering Thoughts on Genetic Enthusiasm’ (1997) 27 Seton Hall Law Review 893, 894

¹⁹² Edward J. Larson, ‘Confronting Scientific Authority with Religious Values: Eugenics in American History’, in Timothy J. Demy and Gary P. Stewards (eds), *Genetic Engineering: A Christian Response- Crucial Considerations in Shaping Life* 105, 106 (Kregal Publications 1999) See also Morse Hyun-Myung Tan, ‘Advancing Civil Rights, the Next Generation: The Genetic Information NonDiscrimination Act of 2008 and Beyond’ (2009) 19 Health Matrix 63

¹⁹³ *Buck v Bell* 274 U.S. 200 (1927)

¹⁹⁴ Daniel J. Kevles, *In the Name of Eugenics: Genetics and the Uses of Human Heredity* (Harvard University Press 1985) 75

¹⁹⁵ Kenneth L. Garver and Bettylee Garver, ‘The Human Genome Project and Eugenic Concerns’ (1994) 54 American Journal of Human Genetics 148, 149

¹⁹⁶ Gordon R. Mitchell and Kelly Happe, ‘Informed Consent after the Human Genome Project’ (2001) 4 Rhetoric and Public Affairs Number 3 375, 379

¹⁹⁷ Sickle Cell Anemia Control Act 1972 Public Law No. 92-294

genetic knowledge can be incorporated into oppressive state policy to dispossess individuals of fundamental human rights.

8.5 Eugenics and the new genetic era

By examining the historical experience of eugenics in Europe and the US, it provides a backdrop to an examination of the potential for eugenics in this new genetic age. With genetic science rapidly advancing and the benefits of science being realised, eugenics considerations are once again coming to the fore.

With this age of “*new genetics*”, the ethical, legal and social implications of genetic technology have become more intricate, however, the controversies and societal risks it raises are the same.¹⁹⁸ These new genetic technologies have the potential to idealise the perfect person, designer babies and a population based upon genetic desirability.¹⁹⁹ This potential genetic underclass raises serious ethical and public policy concerns.²⁰⁰ The potential for this new age of genetics and eugenics gives rise to serious public policy concerns, and signals a need to anticipate the creation of a genetic underclass in society and address such concerns.

9. Conclusion

In looking towards the regulation of genetic information, it is necessary to be aware of the ethical issues and public policy concerns which shape the debate, and form the basis for acknowledging competing rights and interests. In addition, as science progresses, new ethical issues, as well as challenging public policy issues arise and are likely to arise in the future. It is therefore necessary for the law to address the ever emerging ethical considerations and changing socio-political needs instead of responding simply to or reacting to change itself, particularly in an age of constantly evolving genetic science.²⁰¹

The key point highlighted in this chapter is the tension or conflict of rights that arises. When we consider the ethical challenges arising in this debate, and in particular, on exploring the interests and potential rights in genetic information, we view this from the perspective of the individual, the family, third parties and society. To varying degrees and for differing reasons, these four groups have an interest in genetic information. A tension is produced between the competing rights, creating a potentially irreconcilable battlefield of rights and leaving many

¹⁹⁸ See generally Troy Duster, *Backdoor to Eugenics* (2nd ed, Routledge, Chapman and Hall 2003)

¹⁹⁹ Glenn E. McGee, *The Perfect Baby: A Pragmatic Approach to Genetics* (Rowman and Littlefield Publishers 1997) 111

²⁰⁰ Colin S. Diver and Jane Maslow Cohen, ‘Genophobia: What is Wrong with Genetic Discrimination?’ (2000- 2001) 149 *University of Pennsylvania Law Review* 1440, 1447

²⁰¹ Michael J. Flower and Deborah Heath, ‘Micro- Anatomical Politics: Mapping the Human Genome Project’ (1993) 17 *Culture, Medicine and Psychiatry* 27

unanswered questions. It is important to be aware of the various rights and interests in genetic information on the considering appropriate regulation of the area.

In the third party context, this thesis narrows the focus to the employment and insurance context, in consideration of the submission that both are important gateways to active participation in society, and facilitate access to a range of important social goods and services. Third party interest in and use of genetic information in these contexts is therefore particularly worrying.

In addition, complex public policy concerns arise which also raise questions in ascertaining the appropriate application of genetic science. Advances in the scientific community may be stifled, if genetic technologies are misused, or if there is a fear of such technologies being misused. As has been illustrated in this chapter there is also potentially a risk of the creation of a genetic underclass and the practice of genetic cleansing. With advancing science, individuals may increasingly be classified according to their health risk and genetic make up. This underclass may be denied employment and access to other social goods throughout their lives on the basis of perceived health risk.

Society may lose out on the contribution of these individuals, who are in a position to make an active and productive addition to the community, and who are being denied this opportunity, despite their current ability. This may result in the further exclusion and isolation of persons with putative disabilities, (and the elderly), and inhibit their active participation in the community. In addition, this may also lead to a spiral of unemployment, poverty and homelessness, resulting in a myriad of social and economic problems. These dilemmas and potential issues need to be taken into consideration as important contours to this debate, and in acknowledging the potential consequences of failing to react appropriately to advancing science.

In light of the fact that misuse of genetic technologies may result in the exclusion of persons with disabilities and may expose a huge range of putative persons with disabilities, the following chapter will highlight the disability perspective taken in this thesis. It will also explore the application of the social model of disability in this area.

Chapter 4: The interface of the social model of disability with the evolution of genetic science

1. Introduction

As highlighted in previous chapters, advances in genetic science and technology are giving rise to a myriad of ethical and legal dilemmas in relation to access to and use of genetic information, particularly by interested third parties, who often have a financial interest in such information. Although it is acknowledged that misuse of genetic information can take place in a number of third party contexts, this thesis narrows the focus to the contexts of employment and insurance (as highlighted in chapter 3).

This thesis submits that misuse of genetic information in these circumstances exacerbates or heightens the susceptibilities of persons with disabilities (particularly putative disabilities), who are already in a vulnerable position in society and who already may be experiencing exclusion and isolation. As highlighted, genetic testing can potentially detect the onset of future disabilities. In addition, certain attitudes to disability may operate to disable the individual, by imputing a disability to that individual on the basis of a positive genetic test result or on the basis of an undesirable family history of illness. Further, with an ageing population, it is inevitable that more and more individuals will be exposed as having putative disabilities. In light of the evident connection between disability and genetics, and on account of the serious individual and societal implications of abusing genetic technologies, this chapter will focus on the current best thinking on the social construct of disability, and how it can shape the discussion in this area. This thesis acknowledges the discourse and theory surrounding the medical and transhumanist models of disability,¹ however, it focuses on the application of the social model of disability, as is most appropriate in this context.

2. A disability rights approach towards genetic discrimination

In terms of a general framework, this thesis takes a human rights and disability rights approach towards the area of advancing genetic science and technology. It places disability rights and their protection at the forefront of this discussion. This section will examine the core elements and ethos of the social model of disability and will highlight the application in this area.

¹ Gregor Wolbring, 'A disability rights approach to genetic discrimination' in Judit Sandor (ed) *Society and genetic information. Codes and laws in the genetic era* (Central European University Press 2004) 161- 187

2.1 Disability and genetics

Before exploring the social model of disability, it is first necessary to highlight the connection between disability and genetics. Chapter 2 of this thesis sets out the potential of genetic testing and what it can reveal. In view of the fact that genetic testing offers the possibility of detecting the onset and increasingly the potential severity of future disability and disease, genetic technologies provide a new lens through which to view disability. The potential predictive power of these technologies clarifies the concepts of future disability, perceived disability, and imputed or putative disability. It is also observed that perceptions of genetic testing and genetic test results may impact upon individuals' and third parties' view of disability and what it means to have a disability.

As illustrated in chapter 2, it is well established that the extent to which genetic technologies predict the onset of disease or disability is dependent upon a myriad of factors including "*variances in gene expression, accuracy of the test, and the stability of linkage between genetic markers and suspect genes.*"² The technology, although advancing quickly, is still at a relatively early stage. It is also established that, to a certain extent, gene expression depends on whether a condition is a monogenic or a multifactorial disease.³ Although there is still uncertainty in relation to the degree of probability with which a genetic test predicts the onset of disease, some commentators have pointed out that "*despite these known uncertainties and imprecisions, our aversion to disability is so great that people who receive a positive result for a disabling genetic condition may be stigmatized.*"⁴ It is observed that such uncertainty and limited accuracy of genetic testing may impact upon and potentially skew perceptions of disability. It may also exacerbate stigmas.

On examining the connection between disability and genetics, it has further been suggested by some commentators that with rapid advances in genetic technology and the sequencing of the human genome, these developments are changing the way we perceive who is "*normal*" and who is "*disabled*."⁵ Miller asks the question, "*if everyone has genetic conditions that are just waiting to express themselves in the future, isn't everyone truly disabled.*" As nobody has perfect genes, he further observes that "*as we will all have knowledge of the potential genetic disorder that we each harbor, disabled people may no longer remain*

² James P. Evans *et al*, 'The Complexities of Predictive Genetic Testing' (2001) 322 British Medical Journal 1052

³ Neil Holtzman *et al*, 'Predictive Genetic Testing: From Basic Research to Clinical Practice' (1997) 278 Science 602, 604

⁴ Ani Satz and Anita Silvers, 'Disability and Biotechnology', in Thomas Murray and Maxwell Mehlman (eds) *Encyclopedia of Biotechnology: Ethical, Legal and Policy Issues* (Published online 15 October 2000) 173

⁵ Paul Steven Miller, 'Genetic Testing and the Future of Disability Insurance: Thinking about Discrimination in the Genetic Age' (2007) 35 Journal of Law, Medicine and Ethics 47, 50

*stigmatized as “the other” in society.”*⁶ It is observed that not only does this impact upon concepts of disability, but it also can have a profound effect on the notion of personhood and concepts of humanity. Societal stigmas may shift and the notion of disability may further transform and develop to encompass a larger portion of the population including those with genetic predisposition to disease or disability. The existence of an ageing population may further expand the proportion of individuals with genetic predispositions. In light of these additional theoretical considerations, and the impact of genetic technologies on disability, the next section will explore the application of the social model in these circumstances.

3. The social model of disability

This section will address the social model of disability, which was developed as a reaction to the undesirable connotations of and implications of the medical model of disability, as well as in response to the growing awareness of persons with disabilities as rights holders in society. Pursuant to the medical model, disability is seen as a problem inherent in the person, caused by biological defect, disease or other health condition. It is generally viewed as giving rise to the perception of the individual being below- normal and having a low quality of life.⁷ Conversely, the social model of disability views disability primarily as a socially devised problem and with the aim of achieving the full integration of individuals with different abilities into society.

The social model of disability was first pioneered in the late 1960s and 1970s. The theory was advanced in the publications of Vic Finkelstein and Michael Oliver in the 1980s, and Colin Barnes in the 1990s⁸ introducing a paradigm shift in thinking towards disability from a human rights perspective. The key message advocated by the social model is that persons with disabilities are not disabled by inherent defects or other internal sources, but through a variety of different barriers in the social environment.⁹ It is therefore acknowledged that there are different barriers that can be created to disable an individual. Physical barriers can be created in the built environment, for example, an inaccessible public building or an inaccessible workplace.

⁶ Ibid

⁷ For further discussion, see Mark Priestley, ‘Constructions and Creations: Idealism, Materialism and Disability Theory’ (1998) 13 *Disability and Society* 75

⁸ Michael Oliver, *Understanding Disability: From Theory to Practice* (Macmillan Press Ltd 1996); Vic Finkelstein, “We” are not disabled, “you” are, in Susan Gregory and Gillian Hartley (eds) *Constructing Deafness* (Continuum 1990), Colin Barnes, ‘The Social Model of Disability: A Sociological Phenomenon Ignored by Sociologists?’ in Tom Shakespeare (ed) *Disability Reader: Social Science Perspectives* (Continuum 1998)

⁹ Bill Hughes and Kevin Paterson, ‘The Social Model of Disability and the Disappearing Body: Towards a sociology of impairment’ (1997) 12 *Disability and Society* 3 325, 328

As noted, negative third party or societal attitudes can also operate to disable a person. For example, employers may not acknowledge the abilities of disabled people but instead see only the barriers that they encounter on a regular basis as a result of the disability. By focusing on the disability rather than an individual's ability to perform the job, employers may be of the view that persons with disabilities are unsuitable for the position. This is clearly unfair - barriers can often be removed through the provision of accommodations or adjustments to work practices and/or the work environment, in line with the ethos of the social model of disability. Central to the social model of disability is therefore the concept of reasonable accommodation, as a tool to secure and advance the rights of persons with disabilities.¹⁰

This concept therefore adopts a theory that rejects the outdated medical view of disability to seeing disability as a consequence of societal barriers.¹¹ The built environment is generally designed and built for non-disabled individuals, thereby excluding persons with disabilities from actively participating and integrating in society.¹²

The social model of disability provokes a connection with the rights based approach, which is an underlying principle in this thesis. In adopting a rights based approach, the social model views persons with disabilities as valued members of our societies, with rights, who are disabled by the physical and attitudinal barriers within societies that individuals create to exclude and stigmatise them. It has been noted that the social model theory has changed the status of persons with disabilities from passive "*objects of rehabilitation and cure*"¹³ to "*rights holders entitled to make demands on social institutions*."¹⁴ An emphasis on rights "*changes the social status and social understanding of persons with disabilities from powerless recipients of their peers' charity to right-holders capable of making demands on the world*."¹⁵

Viewing disability as a human rights issue requires us to recognise the inherent equality of all individuals, regardless of their abilities or disabilities, and obliges society to remove the attitudinal and physical barriers to achieve equality and

¹⁰ For further discussion, see Anna Lawson, *Disability and Equality Law in Britain: The Role of Reasonable Adjustment* (Hart Publishing 2008)

¹¹ Michael Oliver, *Understanding Disability: From Theory to Practice* (Macmillan Press Ltd 1996)
Bill Hughes and Kevin Paterson, 'The Social Model of Disability and the Disappearing Body: Towards a sociology of impairment' (1997) 12 *Disability and Society* 325, 328

¹³ Chai R. Feldblum, 'Definition of Disability Under Federal Anti-Discrimination Law: What Happened? Why? And What Can We Do About It?' (2000) 21 *Berkeley Journal of Employment and Labor Law* 91, 94

¹⁴ Vlad Perju, 'Impairment, Discrimination, and the Legal Construction of Disability in the European Union and the United States' (2011) 44 *Cornell International Law Journal* 279, 284

¹⁵ Richard K. Scotch, 'Models of Disability and the Americans with Disabilities Act' (2000) 21 *Berkeley Journal of Employment and Labor Law* 213, 216 (quoting Paul C. Higgins, *Making Disability: Exploring the Social Transformation of Human Values* (Charles C Thomas Pub Ltd 1992) 199-200)

inclusion of individuals with disabilities.¹⁶ As will be discussed in chapter 6, the social model paradigm provided a core framework for the CRPD.¹⁷

3.1 Stigma and attitudes towards disability

Having highlighted the rationale of the social model of disability, it is necessary to further highlight the merits of applying and utilising this model to address the regulation of genetic information. It is important to highlight the impact of negative attitudes towards persons with disabilities. Such attitudes can result in stigma and ultimately social exclusion and isolation. This stigma can stretch across various facets of society such as employment, education, political life and social interaction, personal life, as well as an individual's integration into the community.

Pursuant to the medical model of disability, persons with disabilities are defined by their disability and disempowered by the perception that they are ill and in need of a cure. Attitudinal barriers therefore often exacerbate the ethos of the medical model of disability and are contrary to the core rationale of the social model of disability. Society has generally found it challenging to address difference and diversity. This is particularly apparent as regards persons with disabilities and those who are perceived to be disabled.¹⁸ People with disabilities have historically been segregated, isolated and abused, as society has struggled with dealing with who are perceived to be inferior individuals. This leads to a breach of fundamental human rights and entitlements. The societal struggle with difference has left persons with disabilities outside the "*paradigm of humanity*."¹⁹ Wendell argues that the non-disabled portion of society segregate persons with disabilities because "*they represent a threat to the security of their own perceptions of self which are embedded in a culture of perfection and control*."²⁰

It is submitted that this new era of advancing genetic science has the potential to exacerbate these societal attitudes towards persons with disabilities. In addressing the issue of stigma attached to disability in this context, Bickenbach warns that genetic information has the potential to become "*the most profound form of stigmatising labeling people with disability have as yet experienced*."²¹ He postulates that "*human difference, when labeled genetically, opens the door to the most profound forms of stigmatisation*."²²

¹⁶ Michael L. Perlin, "'A Change is Gonna Come': The Implications of the United Nations Convention on the Rights of Persons with Disabilities for the Domestic Practice of Constitutional Mental Disability Law" (2009) 29 Northern Illinois University Law Review 483, 483- 484

¹⁷ United Nations Convention on the Rights of Persons with Disabilities (2006) G. A. Res 61/106 ("CRPD")

¹⁸ Susan Wendell, 'Toward a Feminist Theory of Disability' in Helen Bequaert Holmes and Laura M. Purdy (eds) *Feminist Perspectives in Medical Ethics* (Indiana University Press 1992) 63, 74

¹⁹ Ibid

²⁰ Ibid

²¹ Jerome Bickenbach, 'The Perils of Human Genetics' (1996) 1 *Ethics and Intellectual Disability* 2, 2.

²² Ibid

The increasing availability of genetic technologies and genetic information therefore present new and novel tools to discriminate in areas such as employment, insurance and a variety of commercial settings. Such technologies may ultimately isolate and further segregate persons with disabilities. As highlighted above, advancing genetic science and technology may therefore impact upon third parties' perceptions and attitudes towards disability.

Attitudes towards disability provoke consideration of notions such as imputed disability and perceived disability. Therefore, a disability might be imputed to a person, regardless of the current abilities of the person. In the context of discussing genetic discrimination (and other misuse of genetic information), a third party, for example, an employer may take the view that an individual with a genetic predisposition to a certain condition, has a disability, and may therefore be perceived by the employer as unable to do a particular job. Third parties may therefore impute a disability to an individual or regard an individual as having a disability, on the grounds that an individual merely carries the gene for a particular condition. These perceptions may arise regardless of the individual's current health status and current ability to do the job. Consequently, genetic advances operate to shape perceptions of and attitudes towards disability in society.

Pursuant to negative perceptions, an employer might also perceive an employee as being unable to do a particular job in the future, due to a certain genetic predisposition. Indeed it has been observed that *"attitudes, rather than resource constraints, often create the strongest barriers to employment."*²³ Not only is this negative attitude towards genetic predispositions reflective of the medical model, but it is also in disregard of an individual's current ability to do a job, resulting in discrimination and violation of fundamental rights principles and values, therefore contrary to the social model of disability. The ethos of the social model endorses positive attitudes and a positive response to disability, and proscribes such negative attitudinal barriers, which operate as barriers against the full integration of persons with disabilities.

3.2 Evaluation of the social model in this context

It is submitted that the social model of disability provides the most appropriate framework within which to view the area of genetic science and the connection with disability theory. This model represents current best thinking on disability theory and applies a holistic approach towards recognising and achieving the rights of persons with disabilities. Further, the social construction of disability under this model is broad enough to encompass individuals with putative disabilities or those with genetic predispositions to disability. It also focuses on

²³ Statement by Louise Arbour, U.N. High Commissioner for Human Rights, General Assembly Ad Hoc Committee, 7th Session (27 January 2006)

the actions and intention of third parties, thereby taking the focus away from the individual. The emphasis on a rights based theory, with non- discrimination as a focal point, as well as the promotion of positive attitudes towards disability and persons with disabilities reinforces the relevance of the social model in this debate.

From a regulatory perspective, this model provides a core rights based lens, with the objective of acknowledging societal barriers, both physical and attitudinal, and recognising the equal rights of persons with disabilities. In consideration of the potential for abuse of genetic information and the fundamental human rights at stake, it is observed that such a framework is best suited to address these issues.

4. Conclusion

It has been highlighted that the ethical and legal concerns in this area may have a particularly adverse impact on persons with putative disabilities and persons with genetic predispositions to disabilities. This chapter proposes that the social model of disability provides the most suitable framework from which to view and address the challenging issues that arise at the intersection of advancing science, ethics and the law. With a focus on the societal, attitudinal and other external barriers that disable an individual, it provides a core human rights based model and concentrates on acknowledging and achieving the rights of persons with disabilities. In the context of addressing the misuse of genetic information, this model takes the focus away from the individual and places it on the actions and attitudes of third parties.

In the context of addressing the use of genetic information and the issue of genetic predisposition to disability, this model effectively addresses stigma and negative attitudes and how this might operate to disadvantage the individual.

Having set out the scientific framework shaping this debate, the disability perspective taken and having elucidated the conflict of competing rights, a clear need arises to control the use of genetic information. Against the backdrop of the social model of disability and its application in this area, the next part of this thesis will focus on the choice of regulatory frameworks and the international and comparative benchmarks in this area.

PART 2: The Imperative for Regulation and the Choice of Regulatory

Frameworks

Part 1 of this thesis examined the potential for advancing science and technology to reform health care and medical practice, as well as the corresponding potential to raise a range of ethical and legal dilemmas. The focus of these issues was narrowed to the employment and insurance contexts and the area was examined from a disability perspective. The speed at which science is advancing, together with the potential for abuse and the fundamental human rights at stake, point to the need for a clear regulatory regime to address this area. Part 2 contains three chapters and focuses on the regulation of genetic information. It will examine the preferable mode of regulation and choice of regulatory frameworks in legal theory. It will examine the international human rights position, highlighting the imperative for a regulatory regime and the moral impulse for action in this area. Part 2 will also look at the evolving comparative law benchmarks in this area, and highlight the choice of regulatory frameworks adopted in the comparative jurisdictions.

Chapter 5: Mode of regulation and choice of regulatory frameworks in legal theory

1. Introduction

This chapter focuses on the regulation of genetic information. The first part will consider the mode of regulation. It will examine the merits of adopting a stand-alone approach which singles out genetic information in discrete legislation and segregates the issues. This provokes consideration of the concept of genetic exceptionalism, which will be discussed. This section will also consider hard and soft law options, and what is the most appropriate method in this area.

The second part of this chapter will outline the key regulatory frameworks considered in this thesis. As highlighted in chapter 4, a key element to this thesis is the disability approach taken to address the concept of genetic discrimination and to shape the regulatory approach taken to this issue. In terms of regulatory frameworks, this thesis relies firstly on the non-discrimination theory. The reasons for relying on non-discrimination centre primarily on the observation that the non-discrimination approach has been the traditional tool used to fulfill and protect the rights of persons with disabilities. The objective of the non-discrimination approach in these circumstances is to protect against the discriminatory use of genetic information.

This thesis also relies on the privacy theory. It is noted that the analysis and application of the privacy theory is focused on individual privacy as opposed to the concept of group or family privacy (as is explained in chapter 3 which notes the familial nature of genetic information). The main reason behind reliance on the privacy theory stems from the shortcomings with the non-discrimination approach, the particularly sensitive nature of genetic information, and the need for additional protections for genetic information. In this regard, this thesis considers the merits of a dual approach to addressing genetic information encompassing both non-discrimination and privacy theories.

The third regulatory theory considered in this thesis is the property model. The property model is examined as an alternative model for addressing this area primarily in consideration of the sensitive and private nature of genetic information and in light of the traditionally strong and robust protections offered by the property model. In addition, consideration of the application of the property has been generating much attention and discussion recently, particularly with reference to the recent United States Supreme Court case involving Myriad Genetics Inc. In this regard, it was felt that the property model is a topical model through which to explore in the context of this discussion. This chapter will

conclude with an analysis and evaluation of the relative merits and shortcomings of the various regulatory approaches.

It is noted that this thesis does not consider additional categories or theories of regulation. While it is acknowledged that there are other frameworks that may be applied in this field, for example, the concept of bodily integrity (which is certainly appealing in light of the inherently personal nature of genetic information), this thesis maintains a focus on non- discrimination, privacy and property. In this regard, it is submitted that the discussion and analysis of the three regulatory theories of non- discrimination, privacy and property is sufficient to provide a varied, informed and comprehensive theoretical approach towards the regulation of genetic information for the purposes of this thesis.

2. Mode of regulation: a stand- alone approach?

This section will consider the merits of adopting a stand- alone approach that singles out genetic information in discrete legislation (for example, a new EU level directive specifically tailored to this issue), or whether it would be more appropriate to incorporate genetic information under an existing mainstream approach. Such an option would include amending existing legislation or regulating genetic information alongside for example, health information. It is acknowledged that it may be easier, both legally and politically, to amend existing legislation. However, it may not be the appropriate response in this area.

This provokes consideration of the concept of ‘genetic exceptionalism,’ of which there are arguments for and against. This concept focuses on whether genetic information deserves special protection,¹ or whether protection should have a broader application and, for example incorporate all types of health information (including genetic and non- genetic information). Genetic exceptionalism is based on the concept that genetic information is special, has unique characteristics and therefore merits special protection. It is also based on the premise that genetic information gives rise to distinct ethical and legal issues, and misuse of genetic information has particularly serious consequences.

2.1 Genetic exceptionalism

The concept of “*genetic exceptionalism*,” was pioneered by Thomas Murray, who referenced the previous concept of “*HIV Exceptionalism*,” which viewed HIV as

¹ See generally Thomas H. Murray ‘Genetic Exceptionalism and “Future Diaries”: Is Genetic Information Different from Other Medical Information?’ in Mark A Rothstein (ed) *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic* (Yale University Press 1997). See also George J. Annas, ‘Genetic Privacy: There Ought to be a Law’ (1999) 4 Texas Review of Law and Policy 9, 11. For further discussion see Mark A. Rothstein, ‘Genetic Exceptionalism and Legislative Pragmatism’, (2007) Journal of Law, Medicine and Ethics 59; Colin S. Diver and Jane M. Cohen, ‘Genophobia: What is Wrong with Genetic Discrimination?’ (2000- 2001) 149 University of Pennsylvania Law Review 1439

exceptional based on the potential for discrimination and stigmatisation in society.² The theory of exceptionalism has now shifted to the discussion of addressing genetic information.

The term genetic exceptionalism has been described as “*the (societal) practice of treating genetic data as different from other types of health data for the purposes of assessing privacy and security protections.*”³ Against the backdrop of advancing genetic technology, such information may be viewed as deserving of special non- discrimination and privacy protections in light of the fact that it is inherently unique. There are a variety of reasons given to support this view.

Firstly, in light of its sensitive nature, individuals would naturally desire a particular level of protection over this information. It uniquely identifies an individual, and such information can reveal a myriad of personal details, including current and (potential) future health status, physical appearance, as well as behavioural and other traits. Individuals generally expect a certain level of control and protection over such personal information. In addition, genetic information reveals sensitive information, not only about an individual, but also about that individual's relatives, and is relevant for spouses and partners.⁴ The familial nature of genetic information gives rise to additional considerations in relation to privacy, reproduction, and various ethical considerations such as the right to know and the right not to know. Another unique element of genetic information relates to the fact that it is immutable, in other words, we cannot control the genes that we inherit.⁵

At the forefront of this discussion is the fact that genetic information may be predictive of future disease and disability. It has been described as an individual's “*future diary*” as it effectively “*informs our younger selves about our aging selves.*”⁶ The predictive nature of genetic information is a powerful one and one which may be attractive in a variety of different contexts, including third party contexts. The predictive nature of genetic information is unique, both from an individual's perspective in empowering more efficient healthcare choices, and

² Thomas H. Murray, ‘Genetic Exceptionalism and ‘Future Diaries’: Is Genetic Information Different from Other Medical Information?’ in Mark A Rothstein (ed) *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (Yale University Press 1997). See also Ronald Bayer, ‘Public Health Policy and the AIDs Epidemic: An End to HIV Exceptionalism?’ (1991) 324 *New England Journal of Medicine* 1500, 1500-1504

³ Lawrence O. Gostin and James G. Hodge Jr., ‘Genetic Privacy and the Law: An End to Genetics Exceptionalism’ (1999) 40 *Jurimetrics Journal* 21, 31

⁴ *Ibid* at 35

⁵ Sonia Suter, ‘The Allure and Peril of Genetic Exceptionalism: Do we need Genetic Specific Legislation?’ (2001) 79 *Washington University Law Quarterly* 3 669, 706

⁶ George J. Annas, ‘Genetic Privacy: There Ought to be a Law’ (1999) 4 *Texas Review of Law and Policy* 9, 11. He does however clarify that “*it is coded and probabilistic: we are not necessarily going to get every disease that we are genetically predisposed to develop.*” Annas uses this analogy from William Safire's explanation of diaries, which “*inform our aging selves about our younger selves.*” William Safire, ‘Sleazy Senate Inquiry, *New York Times* (New York 25 October 1993) at 19.

from a third party's perspective as representing a useful mechanism for financial objectives.

In addition, it is noted that the public arguably regards genetic science and genetic information as unique. A lack of education and lack of public knowledge surrounding the reality of advancing science and genetics gives genetic information a mysterious and unknown public perception. These perceptions of genetics are enhanced by the media and in cinema.⁷

Another reason in support of genetic exceptionalism relates to the stigma associated with genetic information. In this regard, the misuse of genetic information has led to discrimination, eugenics and in certain circumstances, has exacerbated racism. Indeed this point is reflected in the historical evidence of eugenics, as discussed in chapter 3. This argument is based on the premise that in light of this stigma attached to genetic predisposition to disease and genetic conditions, discrimination or other misuse of genetic information should be specifically prohibited. In the absence of sufficient protections, this may lead to the practice of genetic cleansing and the potential relegation of what are deemed to be genetically undesirable individuals.⁸

A similar argument relates to the evident link between genetic conditions and certain racial and ethnic groups, which may lead to stigma and perpetuate further discrimination and marginalisation amongst certain groups.⁹ This issue has been highlighted in chapter 3. Misuse of genetic information may bring about the segregation and adverse treatment of these already marginalised groups in society.¹⁰

Another reason in favour of singling out genetic information in the regulation context relates to the expressive value of the law, and recognises the valuable protection of a stand-alone, specific regulatory approach. This argument acknowledges the merit of a specific body of law acting as a tool to engrain a strong moral message in society that a certain practice or behaviour is wrong and will not be tolerated. It is acknowledged that a genetic specific approach would contribute towards establishing and engraining a societal and political attitude that misuse of genetic information is wrong and should not be permitted. Establishing such norms at a relatively early stage seeks to ensure that

⁷ Suter refers to the influence of the media in the public perception of genetics. Sonia Suter, 'The Allure and Peril of Genetics Exceptionalism: Do we need Special Genetics Legislation?' (2001) 79 Washington University Law Quarterly 3 669, 678

⁸ Deborah Hellman, 'What Makes Genetic Discrimination Exceptional?' in Verna Gehring (ed) *Genetic Prospects: Essays on Biotechnology, Ethics and Public Policy* (Rowman and Littlefield 2003) 90- 91

⁹ Lawrence O. Gostin, 'Genetic Discrimination: The Use of Genetically Based Diagnostic and Prognostic Tests by Employers and Insurers' (1991) 17 American Journal of Law and Medicine 1 & 2, 109, 111

¹⁰ Janet L. Dolgin, 'Personhood, Discrimination and the New Genetics' (2000- 2001) 66 Brook Law Review 755, 786- 787

fundamental human rights and their protection are placed at the forefront of this debate.

It is acknowledged that each of the above arguments, taken individually, may not necessarily sufficiently differentiate genetic information from other types of personal or health information.¹¹ However, it is submitted that collectively, these arguments and the elements of genetic information combine to shape its unique nature.¹² In particular, the sensitive and predictive nature of genetic information and the vast amount of personal details it can reveal, together with its stigmatising effect and the potential for misuse and breach of fundamental human rights combine to formulate a strong argument in favour of a genetic specific approach towards the regulation of this area. As will be illustrated in chapter 7, the US has advocated in favour of genetic specific legislation. Some EU Member States have also taken the approach of introducing genetic specific legislation (as will be discussed in chapter 10). In another context, at EU level, the ground of race has been segregated in a discrete directive.¹³

2.2 Opposition to genetic exceptionalism

It is also necessary to examine the arguments against a genetic exceptionalism approach. The main arguments in opposition of this theory relate to the proposition that genetic information is not materially different from other health information. Both types of information are sensitive, reveal personal and intimate details about an individual and are of value to third parties. In practical terms, another issue relates to the difficulty in segregating genetic from non- genetic information, in view of the fact that many diseases and conditions have some genetic element. In addition, medical records generally do not segregate genetic from non- genetic information, thereby creating further practical challenges.

In this regard, Rothstein refers to the wider societal issue of access to health care and advocates in favour of legislating for protection of health information as opposed to specific genetic information.¹⁴ This is a particularly strong argument in the US, as it grapples with health care issues. Similarly, Suter asserts that genetic information is not unique and that concerns about abuses of information should not be limited to genetic information but should extend to other medical information.¹⁵ Lemmens makes similar arguments, particularly in the context of

¹¹ Leslie P. Francis, 'You are Born with your Genes: Justice and Protection against Discrimination in the Use of Genetic Information' (2010) 77 Mount Sinai Journal of Medicine 2 188, 190

¹² Ibid

¹³ Council Directive 2000/43/EC of 29 June 2000 implementing the principle of equal treatment between persons irrespective of racial or ethnic origin [2000] OJ L 180/22

¹⁴ Mark A. Rothstein, "Genetic Exceptionalism and Legislative Pragmatism" (2005) *Hastings Center Report*, No 4 27-33.

¹⁵ Sonia Suter, 'The Allure and Peril of Genetics Exceptionalism: Do we need Special Genetics Legislation?' (2001) 79 Washington University Law Quarterly 3 669

health care in the US.¹⁶ Accordingly, such arguments might carry more weight in a jurisdiction like the US, where there are particular problems with health care and access to health care.

Another argument against a genetic specific approach is that genetic specific laws potentially exacerbate and highlight the stigma of genetic conditions by categorising them differently from non- genetic conditions.¹⁷ Indeed, in this regard, it may be the case that as science and technology advance and genetic information becomes more available and more routine in medical care, the stigma attached to some genetic conditions may be less apparent.¹⁸

2.3 The appropriate mode of regulation

In the context of legislative initiatives, although the arguments against this approach are recognised, on balance, it is submitted that a stand- alone, genetic specific approach is preferable. It would highlight the importance of protecting genetic information, highlight the unique characteristics of genetic information and acknowledge the potential for misuse. It would also potentially help to alleviate the fear of discrimination and other misuse of genetic information by enhancing public confidence. This would have the result of promoting genetic research and innovation and allowing science to flourish. It would have the result of improving the application of genetic science and technology in medical practice, with health benefits for all.¹⁹ There is an overall societal interest in ensuring that genetic science progresses, without interference, and this may well be achieved (to a certain degree) by the adoption of genetic- specific legislation. A specific stand- alone approach would also ensure a degree of clarity and certainty and offer the opportunity to tailor a regulatory response to the particular issues arising.²⁰ It is noted that the option of amending existing legislation would not have the same impact in this area as introducing such a specifically tailored regulatory regime.

It would also carry an expressive value and engender a message that misuse of genetic information is morally reprehensible. For future generations it is important to establish a framework for protecting genetic information and upholding fundamental human rights. Accordingly, this thesis will consider a genetic specific approach at EU level. Having highlighted the merits of adopting a genetic specific

¹⁶ Trudo Lemmens, 'Selective Justice, Genetic Discrimination and Insurance: Should we Single out Genes in our Laws?' (2000) 45 McGill Law Journal 347, 349

¹⁷ Mark A. Rothstein, "Genetic Exceptionalism and Legislative Pragmatism" (2005) *Hastings Center Report* No 4 27-33. See also Lawrence O. Gostin and James G. Hodge Jr., 'Genetic Privacy and the Law: An End to Genetics Exceptionalism' (1999) 40 *Jurimetrics* 21, 33

¹⁸ *Ibid*

¹⁹ Iulia V. Motoc, 'The International Law of Genetic Discrimination: The Power of Never Again,' in Therese Murphy (ed) *New Technologies and Human Rights* (Oxford University Press 2009)

²⁰ Janneke H. Gerards and Heleen L. Janssen, 'Regulation of Genetic and Other Health Information in a Comparative Perspective' (2006) 13 *European Journal of Health Law* 339, 396

approach, the following section will further examine the appropriate mode of regulation.

3. Hard law or soft law?

This section will consider whether it is preferable to pursue a soft law or a hard law approach to this issue. Hard law indicates legally binding, enforceable instruments. These instruments are reflective of strong legal authority, and which imposes certain binding obligations and responsibilities. Such legal mechanisms have a significant expressive value and send a strong message at a political and societal level. It is a strong means to address and regulate an area, and an effective means of protecting and upholding individual rights. The advantages of this mechanism are clear.

In comparison, soft law is generally not legally binding, and it tends to lack features such as strong enforcement, and penalties. They constitute formally non-binding instruments that do not provide enforceable laws, but rather set a certain standard, with the aim of encouraging compliance and developing norms.²¹ Although not directly enforceable, it is acknowledged that soft law has a certain degree of prescriptivity and may be used to interpret binding instruments. Snyder describes soft law as “*rules of conduct which in principle have no legally binding force but which nevertheless may have practical effect.*”²² Soft law regulation is increasingly used in the areas of biotechnology and science disciplines, particularly in light of the flexible nature of this approach, where the advantages of such approach are clear.²³ It may be easier to adapt to changing circumstances and respond more effectively to new issues that may arise in an area. Soft law may also act as an important first step towards the introduction of hard law.

Soft law has frequently been adopted in the insurance industry, in the form of a moratorium. The moratorium approach (a form of self-regulation) involves a voluntary agreement, for example, not to request genetic testing of insurance applicants or to use genetic test results for a certain period of time.²⁴ There are different types of moratoria including limited amount moratoria and partial moratoria.²⁵

²¹ Alina Kaczorowska, *European Union Law* (2nd edn, Routledge 2011) 90. See also, Linda Senden, *Soft Law in European Community Law* (Hart Publishing 2004)

²² Francis Snyder, ‘The Effectiveness of European Community Law: Institutions, Processes, Tools and Techniques’ (1993) 56 *The Modern Law Review* 1 19, 35

²³ Tony McGleenan, ‘Legal Regulation of Genetic Technology in the United Kingdom: Hard or Soft Options’ in Stefan Muller, Jurgen Simon, Jan W. Westing (eds) *Interdisciplinary Approaches to Gene Therapy: Legal, Ethical and Scientific Aspects* (Springer Berlin Heidelberg 1997) 99- 116

²⁴ Linda Selden, ‘Soft Law, Self- Regulation and Co- Regulation in European Law – Where do they meet?’ (2005) 9.1 *Electronic Journal of Comparative Law* 1 (<http://www.ejcl.org/91/art91-3.html>)

²⁵ Margaret Otłowski *et al*, ‘Genetic Discrimination: International Perspectives’ (2012) 13 *Annual Review of Human Genetics* 433, 446

This approach has been adopted in several European countries as a means to address the use of genetic testing and genetic information in the insurance industry, for example in the United Kingdom (the details of which will be discussed in chapter 10). There are valid arguments to the effect that advances in science and technology require a flexible solution, and one which will take account of inevitable future changes and further scientific development. There is certainly merit to the argument that in light of the fact that this area is still at a relatively early stage of development that it is too soon to enact hard law regulation that might quickly become redundant and outdated. Such an approach is also relatively economic and efficient to introduce.

However, there are obvious disadvantages with soft law approaches. In particular, such an approach may cause problems with compliance and lack strong enforcement mechanisms. Such shortcomings may impact upon the credibility of this approach. Also, soft law regulation can be seen as a mechanism *“whereby certain powerful groups avoid accountability through the conventional legal process.”*²⁶ In this regard, such an approach may be attractive to insurance companies and other third parties, who generally want minimal intrusion and interference with current practices. In addition, there may be question marks over transparency, particularly in a situation where policymaking and implementation are under the control of the same body.²⁷

In contrast to hard law, soft law does not carry the same strong expressive weight and symbolic value, and arguably, on a political and societal level, it may not be taken seriously as a means of regulation (as hard law instruments are). It is also observed that the mechanism of the moratorium, for example, is generally a temporary measure and not a long- term solution to this problem.

On balance, although soft law approaches certainly have merit and may represent a credible and appropriate short-term solution, it is not an ideal means of regulating use of genetic information (in the insurance or the employment context). This form of regulation might not operate in favour of the interests of individuals, particularly in light of the weak enforcement mechanisms and the uncertainty in relation to industry compliance with the provisions. Further, individuals may not feel confident that such an approach fully protects their rights. It is acknowledged however that such soft law regulation may provide an adequate temporary solution, while a longer term solution is being considered.

It is submitted that the appropriate long-term solution is the adoption of a hard law approach, and one that is specifically tailored to genetic information. The following section will now look the choice of regulatory frameworks and the merits and shortcomings of these approaches.

²⁶ Tony McGleenan and Urban Wiesling, ‘Insurance and Genetics: European Policy Options’ (2000) 7 European Journal of Health Law 367, 372

²⁷ Ibid at 372

4. Regulatory Frameworks

This section will explore the choice of regulatory frameworks from which to address this area. Against the backdrop of a clear disability rights approach to this area (with a focus on the social model of disability) and acknowledging the importance of upholding fundamental human rights, there are three main regulatory approaches that merit consideration: the privacy approach, the non-discrimination approach and the property approach. The privacy approach aims to protect access to and disclosure of an individual's genetic information, and a non-discrimination approach aims to prevent the discriminatory use of genetic information. With a focus on control, the property approach seeks to acknowledge and protect ownership rights in one's genetic information. The first section will examine the non-discrimination framework.

5. Non-discrimination model

This section will highlight the rationale and the merits of the non-discrimination approach. It will apply this model to the issue of controlling genetic information and the need to protect against the discriminatory use of such information. In adopting a disability approach to this area, it is noted that the main tool traditionally used has been the non-discrimination tool as a means of acknowledging rights, with a view to recognising persons with disabilities as equal rights holders. This method is a well established and an effective means of addressing and ensuring that fundamental human rights are upheld. For example, the Employment Equality Directive²⁸ has successfully used the non-discrimination mechanism to prohibit discrimination on a range of grounds, including disability. In the US, the non-discrimination framework has also been adopted in a range of contexts, including employment.²⁹ At an international level, one of the main tools used in the CRPD to recognise and secure the rights of persons with disabilities is non-discrimination.³⁰

5.1 The rationale of non-discrimination

This section will highlight the underlying objectives of the non-discrimination framework. Discrimination has been described as *“the denial of equal treatment or the rejection of the equal worth of a person due to one or more characteristics*

²⁸ Council Directive 2000/78/EC of 27 November 2000 establishing a general framework for equal treatment in employment and occupation [2000] OJ L 303/16 (“Framework Directive”) (discussed in chapter 9). For further discussion see Evelyn Ellis and Philippa Watson, *EU Anti-Discrimination Law* (2nd edn, Oxford University Press 2013). See also Dagmar Shiek and Anna Lawson (eds) *European Union Non-Discrimination Law and Intersectionality* (Ashgate Publishing Ltd 2011)

²⁹ Genetic Information Nondiscrimination Act of 2008, Pub. L. No. 110-233, 122 Stat. 881 (2008)

³⁰ CRPD

he or she possesses or is thought to possess.”³¹ In recognising the core human rights element, Hendriks links non- discrimination to the fundamental concept of human dignity.³² The main underlying basis for the concept of discrimination is that it is comparative. This reflects the Aristotelian notion of equality pursuant to which “*things that are alike should be treated alike and things that are unlike should be treated unlike in proportion to their unalikehood*.”³³ A key feature of the non- discrimination principle is therefore that there must be a comparator. An individual will only be found to have been discriminated against when there is a relevant comparable individual (or group), by which the standard can be assessed.³⁴ The comparator in question should be in a comparable position to the individual and the key difference between them should be the discriminatory ground. The general rationale of the non- discrimination principle provides that no individual should experience less favourable or unfair treatment on the grounds of personal traits or characteristics.³⁵

Discrimination as a concept can generally be categorised as both direct discrimination and indirect discrimination. The underlying concept of direct discrimination provides that individuals who are similarly or comparably situated should be treated as such and should not be treated less favourably or less fairly on the grounds of particular traits, characteristics or other grounds, such as disability or genetic information. The concept of direct discrimination is a key element of EU non- discrimination law, and is a defined concept in all of the non- discrimination Directives, which will be discussed in chapter 9.³⁶ Direct discrimination is focused on the individual in question and reflects the Aristotelian principle that things that are like should be treated alike.

Indirect discrimination occurs where a particular requirement, practice, provision or treatment, which appears to be neutrally applied, has a particular disadvantage or a disproportionately adverse effect on a particular group or population (for example, race or ethnic group), and such requirement, practice, provision or treatment cannot be justified. The concept of indirect discrimination is also firmly established in EU law. For example, the Employment Equality Directive, the Race Directive and the Gender Equal Treatment Directive all

³¹ Aart Hendriks, ‘The UN Disability Convention and (Multiple) Discrimination: Should EU Non- Discrimination Law be Modelled Accordingly?’ in Gerard Quinn and Lisa Waddington (eds) 2 *European Yearbook of Disability Law* (Intersentia 2010) 10

³² Aart Hendriks, ‘Protection against Genetic Discrimination and the Biomedicine Convention’ in Jan K.M. Gevers, Ewoud H. Hondius and Joep H. Hubben (eds) *Health Law, Human Rights and the Biomedicine Convention* (Koninklijke Brill N.V., the Netherlands 2005) 210

³³ Aristotle, *Ethica Nicomachea* V.3 1131a- 1131b (W. Ross trans. 1925). See also William Hardie, *Aristotle’s Ethical Theory* (2nd edn, Clarendon Press 1980)

³⁴ Michael P Banton, ‘Discrimination entails comparisons’ in Loenen Titia and PR Rodrigues (eds) *Non Discrimination Law: Comparative Perspectives* (Kluwer 1999)

³⁵ Aart C. Hendriks, ‘Different Definition – Same Problems – One Way Out?’ in Mary Lou Breslin and Silvia Yee (eds) *Disability Rights Law and Policy: International and National Perspectives* (Transnational Publishers 2002) 195

³⁶ For example, Framework Directive, Article 2(a) (see chapter 9)

recognise and prohibit indirect discrimination.³⁷ Therefore, depending on the circumstances, indirect discrimination can be defended if objectively justified by a legitimate aim and the means of achieving that aim are appropriate and necessary.³⁸

The concept of indirect discrimination is important in terms of expanding the scope and effectiveness of the non- discrimination principle. Indirect discrimination effectively moves the focus from the individual to the larger group or minority. This is in contrast to the concept of direct discrimination, the focus of which is on the individual. Therefore, it is observed that the concept of indirect discrimination may operate to address potential stereotypes and stigmas that may be attached to certain groups, certain racial or ethnic minorities, or amongst the sexes.³⁹

As noted, in terms of the objective of non- discrimination, from an individual perspective, non- discrimination law is concerned with addressing unfavourable or unfair treatment. From a societal perspective, non- discrimination law is concerned with addressing and tackling power relations and structural inequities in society.⁴⁰ Pursuant to societal inequalities and divisions, dominant groups may be the main recipients of equality and fundamental human rights, while certain non- dominant and minority groups may face barriers to accessing fundamental human rights and equality.⁴¹ Therefore, the non- discrimination tool seeks to reform these competing power relations and inequalities in society to ensure fundamental human rights and equality for all.⁴²

The objective of non- discrimination law is therefore to provide protection against unfair or unfavourable treatment, from both an individual and a wider societal perspective. Arguably, persons with disabilities are among the groups of persons most in need of and entitled to protection against discrimination. Discrimination against persons with disabilities, putative disabilities and those perceived to have disabilities or genetic predispositions to disability can result in exclusion from society and can operate to inhibit active participation in the community, as well as result in a denial of rights.

Accordingly, it is observed that the non- discrimination tool is an effective mechanism utilised in the protection of persons with disabilities and specifically the protection against discrimination based on the ground of disability. Quinn

³⁷ For example, Framework Directive, Article 2(b) (see chapter 9)

³⁸ Ibid

³⁹ Aart Hendriks, 'The UN Disability Convention and (Multiple) Discrimination: Should EU Non- Discrimination Law be Modelled Accordingly?' in Gerard Quinn and Lisa Waddington (eds) 2 *European Yearbook of Disability Law* (Intersentia 2010) 10

⁴⁰ Ibid

⁴¹ Ibid

⁴² Aart C. Hendriks, 'Different Definition – Same Problems – One Way Out?' in Mary Lou Breslin and Silvia Yee (eds) *Disability Rights Law and Policy: International and National Perspectives* (Transnational Publishers 2002) 195

explores the value of using the non- discrimination tool and analyses its merit in the disability context.⁴³ In illustrating the significance of the non- discrimination model in this context, he considers the various ways in which an individual may be discriminated against, as well as the intentions of the discriminator. He refers to the categorisation of individuals or groups of individuals and how historical perceptions or prejudice may result in the labelling of those who are deemed superior and those who are deemed inferior.⁴⁴ Quinn notes that one of the objectives of the non- discrimination framework is to address these stereotypes and place value on the marginalised or stigmatised individual or group.⁴⁵

Quinn further asserts that one the primary objectives of non- discrimination law in the context of disability is “*to separate fact from fiction – to place a spotlight on the person behind the disability.*”⁴⁶ For example, in the employment field, the objective is to look at the whole person, including the abilities and capabilities of the person, while at the same time, reforming negative attitudes and perceptions about persons with disabilities. The non- discrimination principle (both direct and indirect) helps to address the presumptions of inferiority that have been imputed to persons with disabilities.⁴⁷

It is therefore observed that the non- discrimination approach (as it applies in the context of disability) is reflective of the social model of disability. It addresses negative attitudes that might operate to disable an individual. It focuses on the negative, discriminatory actions of third parties and seeks to prevent such actions and uphold an individual’s right not to be discriminated against. It also addresses the barriers that may be created to inhibit an individual’s full participation in areas such as employment, education and political life. By facilitating an individual’s full and active involvement in the community, as well as social and cultural rights, this has a positive impact on an individual’s dignity by recognising one’s inclusion and role in society. This approach recognises human difference and diversity and aims to prohibit discrimination on the basis of such difference. Having explored the basis of the non- discrimination principle and its rationale, the following section will look at how this might be used in regulating genetic information.

5.2 Non- discrimination to protect genetic information

⁴³ Gerard Quinn, ‘Disability Discrimination Law in the EU’ in Helen Meenan (ed) *Equality Law in an Enlarged European Union – Understanding the Article 13 Directives* (Cambridge University Press 2007) 243- 246

⁴⁴ Ibid at 243- 246

⁴⁵ Ibid at 243- 246. See also Larry Alexander, ‘What makes wrongful Discrimination Wrong; Biases, Preferences, Stereotypes and Proxies’ (1992) 141 University of Pennsylvania Law Review 1 149

⁴⁶ Gerard Quinn, ‘Disability Discrimination Law in the EU’ in Helen Meenan (ed) *Equality Law in an Enlarged European Union – Understanding the Article 13 Directives* (Cambridge University Press 2007) 245

⁴⁷ Ibid

On addressing the issue of genetic discrimination, a non- discrimination approach can operate to prohibit the discriminatory use of genetic information. Reference is made here to the analysis of genetic discrimination in chapter 3. Such discrimination indicates that the individual is treated in a more disadvantageous way than another comparable individual, *“solely or primarily because of his or her genotype or because of a specific genetic defect, without there being a sufficient and reasonable justification for such disadvantageous treatment.”*⁴⁸ Similarly, discrimination might also encompass unfavourable treatment on the basis of perceived disability or perceived genetic defects. In this regard, the non- discrimination tool effectively entails a prohibition on the use of genetic information to single out a specific individual or group for unfavourable treatment. It is observed that both direct and indirect discrimination are potentially applicable.

Direct discrimination may arise where, for example, an employer refuses to hire an individual on the basis that the individual tested positive for the BRCA1 gene, even though the individual is otherwise a suitable candidate for the job. Indirect discrimination may occur where, for example, an apparently neutral employment practice, such as a health and safety screening process that applies to all employees, but may have the effect of highlighting (and potentially eliminating, or otherwise adversely effecting) those with a particular genetic predisposition or ethnic background.

From a societal perspective, the use of a non- discrimination approach to address the regulation of genetic information may also operate to prevent the creation of a genetic underclass and deter the practice of genetic cleansing. It may therefore address and prevent potential societal structures and inequities before they take hold and become more widespread. In this regard, it is noted that the non- discrimination framework has also been used to address inequities on other grounds such as race⁴⁹ and this framework has proved successful in protecting rights in this regard in the EU.

There are certainly clear advantages in adopting a non- discrimination framework in these circumstances. From a practical perspective, it may be the case that an aggrieved individual does not have to prove that an employer, or other third party violated their privacy by accessing and using their genetic information. This may be advantageous in cases where an individual has disclosed his/ her genetic information, and subsequently suffered a violation of rights. In such a case, it

⁴⁸ Janneke Gerards, ‘General Issues Concerning Genetic Information,’ in Janneke H. Gerards, Aalt W. Heringa and Heleen L. Janssen (eds) *Genetic Discrimination and Genetic Privacy in a Comparative Perspective* (Intersentia 2005) 24

⁴⁹ Council Directive 2000/43/EC of 29 June 2000 implementing the principle of equal treatment between persons irrespective of racial or ethnic origin [2000] OJ L 180/22

may be more feasible to demonstrate that the individual has been disadvantaged by way of discrimination rather than by way of a violation of privacy rights.⁵⁰

The non- discrimination tool might also prove effective in circumstances where an employer, insurer or other third party inadvertently comes across details of an individual's genetic information, for example, through a parent's obituary in a newspaper, through other publically available resources, or through casual conversation with an individual. The non- discrimination paradigm prevents the subsequent discriminatory use of genetic information in these circumstances. In comparison, the privacy approach would not be effective to prevent the violation of rights in these circumstances. This is relevant in circumstances where the individual consents to the disclosure of genetic information, for example, where such disclosure may be lawful in the employment context for health and safety purposes.

5.3 Shortcomings of the non-discrimination model

It is also necessary to point out the disadvantages of the non- discrimination approach. The main shortcoming is that this approach requires a comparator, by way of establishing a standard. The non- discrimination principle stipulates that the individual must not only show that he or she has been disadvantaged on the basis of his or her genetic information, but also that other individuals or groups (lacking the relevant genetic flaw or predisposition) have been treated more favourably. It is observed that the comparative requirement of the non- discrimination theory makes it less practical and effective in certain respects, than other models, such as the privacy approach, which does not have the same requirement.⁵¹ This may represent a challenging evidentiary obstacle that claimants may face if bringing such a claim under non- discrimination law. There are evident disadvantages with the non- discrimination approach in this regard.

Before further evaluation of the non- discrimination model as a regulatory option, the following section will examine the privacy and data protection approach.

6. The privacy model (data protection)

This thesis also considers the privacy approach (particularly as it applies through the data protection model) as a key regulatory framework. The concept of privacy ensures security and confidentiality in both information and personal space. Privacy safeguards the dignity and integrity of individuals by protecting that which is considered important and private. Privacy laws generally operate by necessitating consent from the relevant party to access or control his/her genetic

⁵⁰ Janneke Gerards, 'General Issues Concerning Genetic Information,' in Janneke H. Gerards, Aalt W. Heringa and Heleen L. Janssen (eds) *Genetic Discrimination and Genetic Privacy in a Comparative Perspective* (Intersentia 2005) 24

⁵¹ Ibid at 24

information. Third parties can access such information by way of obtaining consent from the individual in question. On the other hand, the individual may refuse to consent in which case the third party cannot access the information.⁵²

The privacy model finds expression through data protection legislation, and it is this model which is particularly relevant in this area. The concept of data protection aims to recognise and protect an individual's right to privacy through controlling the collection, disclosure and use of an individual's personal information.⁵³ By enabling such control over this information, this regime therefore protects individuals against misuse surrounding the collection, use and processing of personal information. This approach is evident in the strong data protection legislation in the EU, which protects personal information, as well as the recent developments in EU data protection law, which intend to extend protection to genetic data (see chapter 9). The protection of personal data and privacy rights have also been recognised and reflected in a number of international human rights instruments. For example, article 22 of the CRPD provides for the right to privacy. The various UNESCO Declarations also refer to the right to privacy throughout, as do the Council of Europe instruments.⁵⁴ Indeed privacy and data protection are also firmly embedded in EU human rights law.⁵⁵ These instruments will be discussed in further detail in chapters 6, 8 and 9.

This section will examine the concept of privacy from various perspectives, before looking at the need to protect the privacy of genetic information. It will address the rationale of the privacy framework, its merits, as well as its shortcomings.

6.1 What is privacy?

Although the focus of this regulatory framework is on the data protection model, this section will highlight the core underlying elements of the concept of privacy and its application in this area. The concept of privacy generally denotes control over personal information and personal space. It has been noted that privacy “*represents control over transactions between person(s) and others, limiting or regulating access to individuals or groups, with the ultimate aim of enhancing autonomy or minimizing vulnerability.*”⁵⁶ It is clear that individuals seek control

⁵² Jessica Roberts, ‘The Genetic Information Nondiscrimination Act as an Antidiscrimination Law’ (2011) 86 Notre Dame Law Review 2 597, 617

⁵³ See generally Paul Lambert, *Data Protection law in Ireland: sources and issues* (Clarus Press 2013) See also Paul Schwartz, ‘European Data Protection Law and Restrictions on International Data Flows’ (1994) 80 Iowa Law Review 471, 471. Marc Rotenberg and David Jacobs, ‘Updating the law of information privacy: the new framework of the European Union’ (2013) 36 Harvard Journal of Law and Policy 2 605, 609- 611

⁵⁴ For further discussion, see *ibid*, at 607- 614

⁵⁵ Charter of Fundamental Rights of the European Union, 7 December 2000, Official Journal of the European Communities, 18 December 2000 (2000/C 364/01, Article 7 and 8

⁵⁶ Mark Taylor, *Genetic Data and the Law – A Critical Perspective on Privacy Protection* (Cambridge 2012) 17. In this context, Taylor refers to the literature and thinking of Margulis – see generally,

over information and situations which are personal and valuable and in the context of this debate, genetic information is “*the most personal information of all.*”⁵⁷ Taking another line of thinking, Laurie focuses on the concept of separateness and identifies two conceptions of privacy, namely spatial privacy and informational privacy.⁵⁸

The concept of spatial privacy looks at privacy as maintaining control over an individual’s psychological or physical self. In circumstances where individuals may be required or forced to undergo genetic testing, this may have a negative and potentially harmful impact upon an individual’s psychological wellbeing, thereby invading that individual’s spatial privacy. Likewise, in circumstances where individuals have already undergone genetic testing and are required to disclose the details of such testing, this may result in similar adverse effects on one’s psychological state. An individual might feel that there is a stigma attached to certain genetic conditions and disclosure of genetic details may cause harm to them. Similarly, these effects may further manifest into physical harms. It is observed that the violation of spatial privacy impacts not only an individual, but also that individual’s family members, who share genetic information, or who have an interest in genetic information.⁵⁹

The concept of informational privacy focuses on an individual’s personal information with a view to controlling access and disclosure, as is reflected in the data protection model. It is clear that individuals generally wish to keep personal information private from outside intrusion, and maintain control over who accesses this information. On consideration of the concepts of spatial and informational privacy, Laurie observes a common thread, which indicates, “*privacy is a state of separateness from others.*”⁶⁰ Similarly, Taylor explains that privacy “*represents a relevant state of separation defined and mediated by particular standards,*” and he refers to the term ‘exclusivity’ in this regard.⁶¹ Therefore, the notion of privacy describes the individual as being physically and psychologically detached or separate from others.⁶² Both spatial and informational privacy are relevant when considering access to and disclosure of genetic information. The notion of separateness reflects an individual’s desire to

Stephen T. Margulis, ‘Conceptions of privacy: current status and next steps’ (1977) 33 *Journal of Social Issues* 3 5, 10. See also, Stephen T. Margulis, ‘Privacy as a social issue and a behavioural concept’ (2003) 9 *Journal of Social Issues* 2 243, 245

⁵⁷ Graeme Laurie, ‘The Most Personal Information of All: An Appraisal of Genetic Privacy in the Shadow of the Human Genome Project’ (1996) 10 *International Journal of Law, Policy and Family* 74-101

⁵⁸ Graeme Laurie, *Genetic Privacy: A Challenge to Medico – Legal Norms* (Cambridge University Press 2002) 6.

⁵⁹ *Ibid* at 6

⁶⁰ *Ibid* at 6. See also Graeme Laurie, ‘Challenging Medical- Legal Norms: The Role of Autonomy, Confidentiality, and Privacy in Protecting Individual and Familial Group Rights in Genetic Information’ (2001) 22 *The Journal of Legal Medicine* 1, 27

⁶¹ Mark Taylor, *Genetic Data and the Law – A Critical Perspective on Privacy Protection* (Cambridge University Press 2012) 25

⁶² *Ibid* at 27

ensure that access to genetic information is controlled, with a view to protecting one's personal, psychological and physical integrity.

Having established the underlying principles of the privacy paradigm, the next section will further look at the reasons for protecting the privacy of genetic information.

6.2 Genetic information and the reasons for protecting privacy

This section will delve deeper into the underlying rationale of protecting privacy. The undesirable implications of violating privacy are clear, and can result in the physical, as well as mental and psychological harm to the individual. Further, the undesirable implications of violating privacy are even more apparent when dealing with genetic information. By compelling disclosure of genetic information or forcing an individual to undergo a genetic test, this may have potentially undesirable implications for that individual's wellbeing, as well as adverse societal consequences.

Laurie further explores the rationale for protecting privacy and offers several reasons to justify privacy protection. Firstly, he refers to the necessity of privacy, to facilitate the development and nurturing of personal relationships.⁶³ Personal relationships and friendships usually require privacy, intimacy and trust in order to develop. Personal information is often shared within these relationships, and respect for such information is generally necessary.⁶⁴

Secondly, the notion of 'separateness' facilitates personal reflection, thereby enhancing personal fulfillment and growth.⁶⁵ It is central to the concepts of dignity and respect for individuals, and privacy facilitates the expression of these fundamental rights. In this regard, privacy operates to respect personhood and an individual's inherent sense of self.⁶⁶ Thirdly, as highlighted above, an individual who is not afforded privacy may suffer harm (both mental and physical). Similarly, unauthorised use or disclosure of personal information can lead to individuals being isolated and segregated from society or potentially becoming victims of violence and discrimination.⁶⁷ These reasons for protecting privacy are arguably amplified when dealing with genetic information.

⁶³ Graeme Laurie, *Genetic Privacy: A Challenge to Medico – Legal Norms* (Cambridge University Press 2002) 7

⁶⁴ Ibid

⁶⁵ Ibid

⁶⁶ Sonia Suter, 'Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy' (2003- 2004) 72 *The George Washington Law Review* 737, 770

⁶⁷ Graeme Laurie, 'Challenging Medical- Legal Norms: The Role of Autonomy, Confidentiality, and Privacy in Protecting Individual and Familial Group Rights in Genetic Information' (2001) 22 *The Journal of Legal Medicine* 1, 28. See also Kent Greenwalt, 'Privacy and Its Legal Protection' (1974) 2 *Hastings Center Study* 45

In the health care context, privacy protections are particularly relevant. The disclosure of medical, and often genetic information is vital to the doctor- patient relationship. In the absence of such disclosure, effective care and treatment of the patient may be challenging. By virtue of the fiduciary relationship between the doctor and the patient, the doctor is obliged to ensure the confidentiality of this information.⁶⁸ This point highlights the connection between privacy and the duty of confidentiality, thereby strengthening the need for privacy protections in the health care setting. Personal information and medical information (including genetic information) are generally regarded as confidential, and the duty of confidentiality provides that such information remains confidential.

This point also highlights the societal interest in privacy. There is a societal interest to protect and to minimise potential harm to individuals. It might also have implications in terms of causing harm to societal values and the public interest of ensuring that core values are protected, without undue interference. Again, this may have particular consequences in the health care context, where the physician- patient relationship of trust is necessary for an effective relationship.⁶⁹ There are further public interest considerations as regards the issue of genetic information. There is a corresponding public interest that genetic science be allowed to advance, without undue interference. This highlights a societal need to respect and protect privacy in these circumstances. If individuals are concerned about the protection of their genetic information, they may be apprehensive about taking advantage of genetic technologies or reluctant to engage in clinical trials for the benefit of science.

As highlighted in chapter 3, privacy has particular relevance in third party relationships, such as in employment and insurance. Personal information can be used to the disadvantage of the individual by interested third parties, such as employers in making decisions concerning the individual. To recap, the disclosure of medical information (including genetic information) leaves the employee particularly vulnerable, because employers may be interested in this information, to make decisions that could negatively affect the employee. Therefore, there is a strong need for trust and privacy in these relationships.

Similarly, in the insurance context, disclosure of medical and genetic information can leave the insured particularly vulnerable because the insurer can make decisions within the relationship based on such information and potentially exclude the individual from insurance pools. There are compelling reasons why an individual would want to protect the privacy of genetic information and maintain control of such information in these circumstances.

⁶⁸ Sonia Suter, 'Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy' (2003- 2004) 72 *The George Washington Law Review* 737, 787

⁶⁹ Graeme Laurie, 'Challenging Medical- Legal Norms: The Role of Autonomy, Confidentiality, and Privacy in Protecting Individual and Familial Group Rights in Genetic Information' (2001) 22 *The Journal of Legal Medicine* 1, 29. See also *X v. Y* [1988] 2 All ER 648

6.2 Shortcomings of the privacy approach

Certain shortcomings with the privacy approach can be pointed out. It is noted that privacy “*does not provide for any continuing protection of or control over personal information and details once they enter the public sphere.*”⁷⁰ This creates the possibility of misuse of this information once it is in the public sphere, perhaps in the form of the discriminatory use of the information. Accordingly, the privacy regime does not protect against the discriminatory use of genetic, (or other personal) information.⁷¹ Similarly, it is observed that a privacy regime alone does not address the potential use of genetic information obtained through other means – for example, through an individual’s inadvertent disclosure of family history or genetic information obtained through publically available sources, such as an obituary.

Another observation can be made of the privacy regime. As has been established, an individual’s genetic information can be very important and a valuable research tool for the scientific community. Moreover, the advances in genetic science have relied upon not only access to genetic information, but also a willingness of individuals to participate in clinical trials and experiments. It can therefore be observed that there may be a certain concern that a strong emphasis or over- reliance on privacy and data protection laws may have an adverse effect on scientific research, and may operate to impede the development of such research.⁷² However, in this regard, it is equally noted that such laws are necessary in order to protect privacy rights in the research context.

It is also observed that as science advances, genetic information will become more appealing to third parties and will become more routinely sought after in application forms and other scenarios. In these circumstances, an individual may be aware of their right to privacy, but in practical terms may feel obliged to reveal such information.⁷³ Individuals may feel that for professional reasons, it is necessary to agree to disclosure. In addition, if individuals refuse to disclose such information, an employer or other third party may nevertheless mistreat the individual, or raise assumptions as to the individual’s perceived genetic profile.⁷⁴

⁷⁰ Graeme Laurie *Genetic Privacy: A Challenge to Medico – Legal Norms* (Cambridge University Press 2002) 300

⁷¹ Janneke Gerards, ‘General Issues concerning Genetic Information’ in Janneke Gerards, Aart Heringa and Heleen Janssen (eds) *Genetic Discrimination and Genetic Privacy in a Comparative Perspective* (Intersentia 2005) 23

⁷² Richard A. Spinello, ‘Property rights in genetic information’ (2004) 6 *Ethics and Technology* 29, 30. See also Bartha Knoppers, EMBO Newsletter, Issue 24 (Summer 2013). Available at: <http://www.embo.org/news/emboencounters/issue-24/the-limits-of-privacy> (accessed 1 August 2013)

⁷³ Jessica Roberts, ‘The Genetic Information Nondiscrimination Act as an Antidiscrimination Law’ (2011) 86 *Notre Dame Law Review* 2 597, 618

⁷⁴ *Ibid*

Before further evaluating this option, the following section will look at the property approach and its merits as a regulatory model.

7. A property law framework

This section will highlight another potential approach from which to regulate genetic information. Property law can provide a strong and effective means to protect important interests, such as one's personal possessions and personal information. Property signifies power and protection and it has been stated that "*property has teeth and symbolic force.*"⁷⁵ It has also been stated that "[t]here is nothing which so generally strikes the imagination, and engages the affection of mankind, as the right of property..."⁷⁶ Property rights can protect both tangible and intangible property.

Property rights encompass a collection of interests or "*bundle of different rights*"⁷⁷ that an individual may have.⁷⁸ It has been further observed that the characteristics of property relating to its use, transfer, and enjoyment are known as '*property interests*' and collectively these interests indicate some form of ownership.⁷⁹ Property interests encompass "*exclusive possession or enjoyment, control over how the object is used or kept from use (transferability), alienability, divisibility, and the length of term of ownership interests.*"⁸⁰ In some ways, these principles are also reflected in the privacy model in terms of ensuring control over one's personal information. It is also observed that the property framework protects and upholds privacy rights and affords individuals control over access to and use of their personal property. In addition, property rights (in one's genetic information) can also act to uphold an individual's autonomy and self-determination.

Two elements are immediately appealing when considering the merits of property rights – it denotes ownership and therefore control.⁸¹ Property rights also signify value and worth. Such language of ownership is often used to refer to important and valuable things, that we wish to maintain control of.⁸² As a legal framework, property offers protection of important interests, it has "*a long legal tradition*" and, therefore it may be viewed as a clear-cut mechanism to protect interests.⁸³ It is

⁷⁵ David F. Partlett 'Misuse of Genetic Information: The Common Law and Professionals' Liability' (2003) 42 Washburn Law Journal 489, 497

⁷⁶ William Blackstone, *Commentaries on the Laws of England* Volume 2 (Oxford 1766)

⁷⁷ James Penner, *The idea of property in law* (Oxford University Press 1997) 1

⁷⁸ Discussions of property often use the words "*right*" and "*interest*" interchangeably. Arthur L. Corbin, 'Legal Analysis and Terminology' (1919) 29 Yale Law Journal 163, 163

⁷⁹ Lawrence C. Becker, *Property Rights: Philosophic Foundations* (Routledge 1980) 18

⁸⁰ Catherine M. Valerio Barrad, 'Genetic Information and Property Theory' (1992- 1993) 87 Northeastern University Law Review 3 1037, 1049

⁸¹ Sonia Suter, 'Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy' (2004) 72 The George Washington Law Review 4 737, 753- 754

⁸² Ibid at 750

⁸³ Ibid

noted that the right to property has also been recognised in a number of European legal instruments. For example, Article 17 of the CFREU provides for a right to property (including intellectual property).⁸⁴ This provision is reflective of Article 1 of the First Protocol to the ECHR.⁸⁵ The ECJ has also recognised the right to property, in the *Hauer* case, and referred to the right to property set out in the First Protocol to the ECHR.⁸⁶

This section will further explore the concept of property. It will address the underlying rationale of the property framework, its merits and its shortcomings. It will look at the potential for creating property rights in genetic information.

7.1 Property rights to protect genetic information

In the context of discussing property rights, it is important to highlight the distinction between tangible and intangible property rights. Ownership rights in this area would confer such rights on genetic material, which would encompass actual genetic material derived from the human body, and the genetic information obtained from such genetic material.⁸⁷ For the purposes of this debate, we are particularly concerned with genetic information, which is an intangible source. It has been observed that “*the value of intangible genetic data about an individual is probably not comparable to the value of an individual’s cells from which a commercial product is derived.*”⁸⁸ Yesley further looks at the distinction between genetic material and genetic information and notes that in light of the fact that genetic information is not seen as having the same commercial value as genetic material, it would not “*warrant the cost of enforcement by those affected.*”⁸⁹ Nevertheless, genetic information is significantly valuable, not only to the individual, but also to third parties, who may be interested in using genetic information as a powerful predictive tool for financial advantage. This information can build up a valuable profile of potential and current employees, insurance applicants and can give these third parties insights into the current and future health and behavioural patterns of such individuals.

The use of property to address genetic information is enticing because generally we use the possessive to refer to something that is important and meaningful.⁹⁰ Genetic information is very personal information revealing sensitive details about

⁸⁴ Charter of Fundamental Rights of the European Union, 7 December 2000, Official Journal of the European Communities, 18 December 2000 (2000/C 364/01), Article 17

⁸⁵ First Protocol to the Convention for the Protection of Human Rights and Fundamental Freedoms CETS No. 009 [1952]

⁸⁶ Case 44/79 *Liselotte Hauer v Land Rheinland-Pfalz* 1979 ECR 3727

⁸⁷ Richard Spinello ‘Property rights in genetic information’ (2004) 6 *Ethics and Technology* 29, 30

⁸⁸ Michael Yesley, ‘Protecting Genetic Difference’ (1998) 13 *Berkeley Technology Law Journal* 653, 664

⁸⁹ *Ibid* at 663

⁸⁹ *Ibid*

⁹⁰ Sonia Suter, ‘Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy’ (2004) 72 *The George Washington Law Review* 4 737, 750

an individual. It is personal information, disclosure of which can potentially cause great harm to the individual. The property rights framework offers strong and comprehensive protection across a broad spectrum. In this context, the main objective of using property to protect genetic information is “*to control its use and disclosure.*”⁹¹ The recognition of property rights in genetic information is appealing in light of the fact that it indicates market control and facilitates “*the ability to buy and sell the object as a commodity.*”⁹² In light of the value of genetic information, such a framework would offer a strong means by which to protect such interests.

Similar arguments have been made in favour of recognising body parts (a source of genetic information) as property, particularly in the research context.⁹³ The denial of property rights may result in the exploitation of the individual whose genetic information may be utilised for economic gain. This argument has merit in situations where researchers profit from an individual’s genetic information or samples.

At first glance, property law therefore seems to be a natural choice for recognising and respecting genetic differences, in light of the value and significance of genetic information and genetic material, and the potential benefits of genetics being realised. Treating genetic information as property also serves an important expressive function. From a societal perspective, it acknowledges that our genetic material is a valuable and unique commodity and something worthwhile protecting. Although the property framework appears to offer an attractive means of providing strong protection of genetic information, certain shortcomings can be identified.

7.2 Shortcomings of the property framework

This section will highlight some of the undesirable aspects of adopting a property rights model. It is observed that one of the main problems with using property rights in this context is the potential for commodification. Commodification explains the process whereby goods and services or other items are transformed into a commodity or commercial product. It has been stated that “[A]ll forms of property...involve some type of commodification, which is a social process in which people treat things ... as property and apply market rhetoric to those things.”⁹⁴

⁹¹ Ibid at 664

⁹² Sonia Suter, ‘Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy’ (2004) 72 The George Washington Law Review 4, 737, 754

⁹³ For further discussion, see E. Richard Gold, ‘Body Parts: Property rights and the ownership of Human Biological Materials’ (1997) 10 Harvard Journal of Law and Technology 2 369

⁹⁴ David B. Resnik, ‘The Commercialization of Human Stem Cells: Ethical and Policy Issues’ (2002) 10 Health Care Analysis 127, 140

Concerns about commodification frequently arise in discussions of property rights in the body. Suter points out that the commodification of things deemed personal can present problems.⁹⁵ First of all, by using economic or market-based language to define an individual's genetic information, there is a potential that this may diminish the inherent value and dehumanise our interest in such information.⁹⁶ As a result, it may have an undesirable effect upon the notion of personhood and an individual's sense of self, as well as fundamental concepts of bodily integrity and autonomy, which are generally not defined in such terms. By impacting upon one's sense of self, it may further devalue personal interests in genetic information, which may have further implications for an individual's relationships and interaction with others.⁹⁷ To a certain extent, commodification of genetic information is already taking place, particularly with reference to the information society which is fast becoming a reality, however, it is submitted that granting property rights would further amplify the commodification process.

The process of commodification as it applies to genetic information, genetic material or other inherent concepts, provokes a concept of self as something that can be segregated, and something that can be bought, sold and can engage in economic activity.⁹⁸ In this regard, commodification is arguably "*incompatible with human dignity*."⁹⁹ Accordingly, it is acknowledged that property law fails to take into account the importance of human rights, particularly rights to dignity and integrity of the person. It also arguably fails to acknowledge the importance of recognising the notion of personhood, raising further ethical concerns in defining humanity.

Linked to the above argument concerning commodification, it is also noted that property law confers a purely economic right, which arguably fails to acknowledge the dignitary concerns underlying the protection of genetic information. Treating genetic information as personal property could violate our humanity by viewing people, not as human beings, but as groups of potentially profitable genetic traits and by putting an actual market value on our individual value and worth.¹⁰⁰ It might encourage those in financially poor situations to reveal or trade personal genetic information with a view to earning a profit. This may potentially create a black market for genetic information and result in the further misuse of genetic information with undesirable ethical and public policy implications.

⁹⁵ Sonia Suter, 'Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy' (2004) 72 The George Washington Law Review 4, 737, 799

⁹⁶ Ibid

⁹⁷ Ibid at 800

⁹⁸ Ibid at 800

⁹⁹ Lars Oystein Ursin, 'Privacy and Property in the Biobank Context' (2010) 22 Healthcare Ethics Committee Forum 211, 221

¹⁰⁰ Jessica Roberts, 'The Genetic Information NonDiscrimination Act as an Antidiscrimination Law' (2011) 86 Notre Dame Law Review 2 597, 612

Another drawback of the property rights framework in this area relates to the concept that genetic information is often shared between individuals, for example, family members, and sometimes larger communities. Accordingly, an individual's actions as regards their genetic information may potentially impact upon others.¹⁰¹ It has been noted that "*the property model undermines the relationships in which we share this information, pushing them toward arms-length transactions as opposed to relationships of trust.*"¹⁰² This is particularly problematic in light of the familial nature of genetic information. This dilemma therefore raises serious ethical issues and raises questions over control and use of familial genetic information.

Another problem with the use of a property rights framework relates to the potential for economic inefficiency, which may consequently have a stifling effect on scientific research.¹⁰³ In this regard, Spinello refers to "*fragmented property rights in the genetic data coming from multiple sources.*"¹⁰⁴ This might result in additional costs and efforts in negotiating various potential interests, addressing potential licensing fees and reaching agreement on the use of such genetic material or information.¹⁰⁵ This has adverse implications in terms of economic efficiency and subsequent scientific advances. Although this argument is particularly relevant in relation to potential property rights in tangible genetic material, it is also a valid consideration as regards intangible genetic information.

The arguments against the recognition of property rights in these circumstances, particularly with respect to commodification are reflected in case law. In *Moore v Regents of the University of California*, the Supreme Court argued that granting Mr Moore property rights in his spleen would amount to making commodification of the human body legitimate.¹⁰⁶ The court also held that conferring property in such genetic material would stifle scientific research and development, and stated that "*this exchange of scientific materials, which is still relatively free and efficient, will surely be compromised if each cell sample becomes the potential subject matter of a lawsuit.*"¹⁰⁷ The case highlights the point that society has a clear interest in advancing scientific research and technological innovation. The existence of any such property rights may act as barriers to and burdens on such research and innovation.

¹⁰¹ Mathew Stulic, 'Genetic Non Discrimination, Privacy and Property Rights' (2000) 7 Murdoch University Electronic Journal of Law 2. Available at: <http://worldlii.austlii.edu.au/au/journals/MurUEJL/2000/19.html> (accessed 25 July 2013)

¹⁰² Sonia Suter, 'Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy' (2004) 72 The George Washington Law Review 4 737, 747

¹⁰³ Richard Spinello 'Property rights in genetic information' (2004) 6 Ethics and Technology 29, 35 (Kluwer Academic Publishers)

¹⁰⁴ Ibid at 35

¹⁰⁵ Ibid

¹⁰⁶ *Moore v Regents of the University of California* (1990) 51 Cal. 3d, 120, 271 Cal. Rptr. 146; 793 P2d. 479

¹⁰⁷ Ibid at para 97

7.2.1 Patenting genes

As noted, property law encompasses both tangible and intangible assets. Pursuant to intellectual property law (which incorporates patents) owners are conferred exclusive rights over intangible assets. A patent is “*a legal right of an inventor to exclude others from making or using a particular invention.*”¹⁰⁸ The patentability of genes and the issues that arise in this regard are also relevant in this debate, in illustrating the commodification of genetic material and genetic information and the other challenges this may raise.¹⁰⁹

In a high profile case that came before the US Federal Court of Appeals, Myriad Genetics Inc sought to patent two genes linked to breast and ovarian cancer, called BRCA1 and BRCA2.¹¹⁰ Those in favour of patenting genes propose that it is vital as a motivation for scientific research and technological endeavours, as was argued by Myriad Genetics Inc. Those against such patenting argue that it is contrary to human dignity and the inherent value of human beings. It might also result in exploitation by scientists and companies who may take financial advantage and control of their dominant commercial position in the market, to the detriment of individuals who's health may benefit from taking these genetic tests. In this case, the Federal Court of Appeals affirmed the right of Myriad Genetics Inc to patent these two genes. The case was then appealed to the Supreme Court. In this landmark case, the Supreme Court recently found that human genes cannot be patented, on account of being products of nature.¹¹¹

Therefore, there are valid policy reasons (as well as strong ethical reasons) against granting property rights in genetic information and genetic material.

8. Evaluation of the various regulatory options

This section will briefly evaluate the various regulatory options. As regards the property model, it is acknowledged that such a framework provides a useful and forceful means of protecting personal genetic information. Property rights can be utilised as an effective means of ensuring control and ownership over genetic information and genetic material. The provision of property rights may ensure that appropriate financial reward is provided to individuals, with the objective of preventing exploitation. These rights can also protect the privacy rights and

¹⁰⁸ Bronwyn H. Hall, 'Patents and Patent Policy' (2007) Oxford Review of Economic Policy 1. See also Robin Feldman, *Rethinking Patent Law* (Harvard University Press 2012)

¹⁰⁹ Sirpa Soini *et al*, 'Patenting and licensing in genetic testing: ethical, legal and social issues' (2008) 16 European Journal of Human Genetics 10

¹¹⁰ *The Association for Molecular Pathology et al v Myriad Genetics, Inc et al* 569 U.S. 12-398 (2013)

¹¹¹ *Ibid*. See also Aaron S. Kesselheim *et al*, 'Gene Patenting – the Supreme Court finally speaks' (2013) New England Journal of Medicine. (Massachusetts Medical 2013) Available online at: <http://www.nejm.org/doi/pdf/10.1056/NEJMhle1308199> (accessed 9 August 2013). For discussion of the European position as regards the patenting of the breast cancer gene, see Gert Matthijs, 'The European opposition against the BRCA gene patents' (2006) 5 Familial Cancer 95

autonomy of individuals whose genetic information is sought for financial self-interest, or for research purposes.

It is observed that the property model appears to be linked to the privacy model to a certain degree. There are certainly links that can be made between the property and the privacy models, particularly in terms of the control offered. However, on closer inspection, it may be the case that property rights in genetic information may in fact operate to harm privacy interests. Property rights may damage the relationship of trust in which genetic information is disclosed, with particular reference to the commodification of genetic information.¹¹²

It is noted that the privacy model requires consent for obtaining and using one's genetic information, giving individuals control over the use of their information. However, this kind of control differs from that of property, and "*captures the way in which disclosure of personal information is a giving of oneself and a form of entrustment... rather than consent to arms-length business transactions.*"¹¹³ In addition, the right to privacy is fundamental and inalienable. This is contrasted with the right to property, the essence of which is alienability. Further, as highlighted, property rights do not necessarily denote respect for dignity and integrity of the person, whereas these are core elements of the privacy framework. Although certain similarities can be identified, the differences between the two frameworks are clear.

On balance, it is submitted that particularly in light of the potential commodification of genetic information and the undesirable consequences with regard to one's dignity and integrity, the property framework is not appropriate as a regulatory framework. Property rights do not necessarily take into account the familial nature of genetic information, as well as the limited right to know of employers, insurance companies and other third parties.

As regards the non-discrimination approach, it is submitted that the merits of this approach far outweigh the few shortcomings, and there are compelling reasons in favour of adopting such a framework in this area. The non-discrimination principle can facilitate enhanced lives for all individuals, and can open the doors to accessing a myriad of fundamental human rights. It is a broad concept and encompasses direct and indirect discrimination. The non-discrimination principle also has a strong expressive value and can operate to engrain a message in society that discrimination on the basis of a broad range of characteristics, such as genetic information, is morally and legally wrong. The non-discrimination tool has been used successfully in various jurisdictions as a means of addressing disability and other discriminatory grounds. However, although this approach offers a high level of protection, the question arises whether a sole non-discrimination approach is sufficient to fully protect genetic information.

¹¹² Sonia Suter, 'Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy' (2004) 72 The George Washington Law Review 4 737, 805

¹¹³ Ibid at 811

The privacy approach, as is expressed in the data protection model also has a lot to offer in terms of a regulatory framework in this area. It is clear that an individual would not wish for genetic information to be freely accessible or available to others in the absence of permission or informed consent. Therefore, individuals would wish to control access to genetic information and how and under what circumstances such information is used. Under the privacy and data protection model, it is acknowledged that a person's genetic information is uniquely personal and private and therefore should be under his/her control. There are compelling reasons why an individual would wish for genetic information to remain private and safe from intrusion.

Such an approach would provide relatively strong protection in terms of control over genetic information. It would also acknowledge the sensitive nature and value of genetic information as well as an individual's dignity and integrity in maintaining control over such personal information. In addition, this framework can operate to uphold and maintain the rights to autonomy and self-determination. Privacy rights effectively respect an individual's right to determine when, and in what manner their genetic information is used. In comparison to one of the key elements of the non- discrimination principle, there is no need for a comparator in order to gain protection, which is appealing from a practical and evidentiary perspective.

In light of the evident shortcomings, it is clear that the privacy framework can only reach so far and there are certain gaps that can be identified that cast doubt as to whether such a regime would operate to fully protect genetic information or instill the necessary confidence in individuals. Similarly, it is submitted that protecting genetic information by way of non- discrimination alone may not be sufficient to fully protect and prevent misuse.¹¹⁴

This raises the question whether some form of a dual privacy (data protection) and non- discrimination approach may be preferable. In light of the unique and sensitive nature of genetic information, as well as the potential for abuse, and in consideration of the inadequacies of the individual regulatory approaches, it is submitted that the pursuit of both a privacy and non- discrimination approach might be the preferable way forward in the regulation of this area.¹¹⁵ It would operate to regulate the access to and disclosure of genetic information, as well as the discriminatory use of such information.

9. Conclusion

¹¹⁴ Pauline T. Kim, 'Genetic Discrimination, Genetic Privacy: Rethinking Employee Protections for a Brave New Workplace' (2002) 96 Northwestern University Law Review 1497, 1498

¹¹⁵ Janneke H. Gerards and Heleen L. Janssen, 'Regulation of Genetic and Other Health Information in a Comparative Perspective' (2006) 13 European Journal of Health Law 339, 397

This chapter focused on addressing the regulation of genetic information. In terms of the mode of regulation, this section also considered the merits of adopting a stand-alone regulatory model in this area that recognises the merits of singling out genetic information as being worthy of special protections, and found that this is the preferable approach in this area. This chapter also asserts that the preferable method to consider is hard law, particularly in light of its binding nature, strong enforcement and effective means of protecting individual rights.

In terms of regulatory frameworks, this chapter addresses the non-discrimination approach, privacy/ data protection approach and the property approach. Although a strong and effective means of regulation and protecting rights, it is concluded that the property model is not an appropriate means of regulating genetic information. It raises serious ethical concerns and it is likely that the utilisation of property rights in this regard may skew perceptions of human beings as objects that can be bought and sold.

This chapter also examined the non-discrimination model, which has had a successful history in protecting rights and has traditionally been used as a means to recognise the rights of persons with disabilities. Its objective in these circumstances is to protect against the discriminatory use of genetic information. It is also acknowledged that non-discrimination is a reliable and well-established principle, which is firmly positioned in international and EU law. Although shortcomings can be identified with this model, it is submitted that this model would provide the most appropriate means of regulating genetic information.

It is also submitted that in order to ameliorate the shortcomings with this approach and to fully protect the rights of individuals, this approach could be supplemented by the privacy/ data protection approach. This mechanism offers strong protection to uphold an individual's rights. The privacy model also has its shortcomings, particularly in light of the lack of protection it offers against discrimination, and it is submitted that as a discrete approach, it would not offer a sufficiently robust or effective regulatory regime in this area.

In terms of the regulation of genetic information, it is concluded that the preferable approach is utilisation of both the non-discrimination approach and the privacy approach, as a two-pronged mechanism for ensuring the full protection of genetic information. Such a framework would ensure that access to and disclosure of genetic information is controlled and would ensure that the discriminatory use of genetic information is prohibited. In consideration of the unique and sensitive nature of genetic information, it is concluded that such a comprehensive legislative framework is preferable and perhaps necessary in order to ensure the protection of fundamental human rights and instill confidence in individuals that genetic technologies can be used to enhance health care without the fear that it will be used to disadvantage.

Having examined and evaluated the available regulatory models and the preferable approach in this area, the following chapters will provide an overview as to the international human rights position and the evolving comparative law benchmarks in this area.

Chapter 6: Framings from International Human Rights Law

1. Introduction

Since the introduction of the Universal Declaration of Human Rights in 1948¹ human rights and their interpretation have continued to evolve, as society has evolved, with the introduction of various treaties and soft law instruments. This chapter will explore the international position in relation to advancing genetic science and the protection of genetic information, from a human rights perspective. The first part of this chapter will provide an overview of the relevant UN treaties which shape the international human rights framework. It will include discussion of the International Covenant on Civil and Political Rights² and the International Covenant on Economic, Social and Cultural Rights.³

There will be a particular focus on the CRPD,⁴ and its influence on this area, from a disability rights perspective. This section will explore the CRPD in facilitating a framework for discussion of these issues, as well as the potential impact it might have on the regulation of genetic information in the EU. In characterising the debate on genetic discrimination, this section will highlight the focus on the non-discrimination framework as a key theme running through the CRPD. This section will also explore the relevance of the conclusion of the CRPD by the EU, and the subsequent obligations and responsibilities on the EU as a result.

This chapter will then focus on international soft law which expresses a concern for the protection of genetic information and indicates a desire for regulatory intervention in this area. In this respect, it will focus on the declarations released by the United Nations Educational, Scientific and Cultural Organisation (UNESCO) which recognises the need for moral standards in the application of science.⁵ The UNESCO has produced three important instruments that are applicable to the debate of protecting genetic information, all of which will be discussed in this chapter. The key instruments are the Universal Declaration on the Human Genome and Human Rights (1997), the International Declaration on Human Genetic Data (2003) and the Universal Declaration on Bioethics and Human Rights (2005).

¹ G.A. Res. 217 (111) of 10 December 1948, UN Doc. A/810 (1948) ("UDHR")

² G.A. Res. 2200A (XXI), 21 UN GAOR Supp. (No. 16) at 52, UN Doc. A/6313 (1966), 999 UNTS 171 (entered into force 23 March 1976) ("ICCPR")

³ G.A. Res. 2200A (XXI), 21 UN GAOR Supp. (No. 16) at 49, UN Doc. A/6316 (1966), 993 UNTS 3 (entered into force 3 January 1976) ("ICESCR")

⁴ United Nations Convention on the Rights of Persons with Disabilities (2006) G. A. Res 61/106 ("CRPD")

⁵ Sirpa Soina, 'Genetic Testing legislation in Western Europe – a fluctuating regulatory target' (2012) 3 Journal of Community Genetics 143, 145

2. General United Nations Human Rights Treaties

The first part of this chapter will give an overview of the relevant human rights treaties of the UN and their value in shaping key fundamental human rights principles. Before discussing the CRPD, this section will highlight the International Covenant on Civil and Political Rights (ICCPR) and the International Covenant on Economic, Social and Cultural Rights (ICESCR), both of which are essential to the international human rights framework.

In 1966 these two treaties were adopted, one with the objective of advancing civil and political rights, and the other with the objective of advancing economic, social and cultural rights. Both of these treaties came into force in 1976. These treaties are based upon core human rights principles and primarily use the non-discrimination approach to recognise these rights. The aim is to promote equality and to outlaw discrimination for all individuals, while acknowledging other basic rights. The following sections will highlight the relevance of these treaties in framing human rights discourse.

2.1 International Covenant on Civil and Political Rights

The ICCPR focuses on the recognition and protection of civil and political rights for individuals.⁶ In mirroring the ethos of the Declaration of Human Rights 1948, the ICCPR adopts a human rights framework to achieve these rights. Article 1 contains the right to self-determination, which provides the right to decide one's political objectives, as well as economic, social and cultural objectives.⁷ There is also a clear non-discrimination approach evident. For example, Article 2 states the ICCPR will be respected and ensured “...*without distinction of any kind such as race, colour, sex, language, religion, political or other opinion, national or social origin, property, birth or other status.*”⁸

Article 2 has been further clarified by General Comment No. 20, which provides, *inter alia* “...*Membership [of a group] also includes association with a group characterized by one of the prohibited grounds (e.g. the parent of a child with a disability) or perception by others that an individual is part of such a group.*”⁹ This provision therefore intends to extend protection to, for example, individuals who are not themselves disabled, but who are discriminated against ‘on the basis of disability.’ In addition, Article 26 provides for equality before the law, and states “*All persons are equal before the law and are entitled without any*

⁶ See generally, Nigel Rodley, ‘Civil and Political Rights’ in Catarina Krause and Martin Scheinin (eds) *International Protection of Human Rights: A Textbook* (2nd edn, Abo Akademi University Institute for Human Rights 2012)

⁷ ICCPR, Article 1

⁸ ICCPR, Article 2

⁹ CESCR General Comment No. 20 (2009) Non-Discrimination in Economic, Social and Cultural Rights, UN Doc E/C.12/GC/20, para 16

*discrimination to the equal protection of the law. In this respect, the law shall prohibit any discrimination ... on any ground such as race... or other status.*¹⁰ This general equality provision provides broad protection across a number of discriminatory grounds.

Although 'disability' is not expressly provided for in Article 2 or Article 26, it is arguable that it could come within the ambit of 'other status.'¹¹ In addition, the Committee on Economic, Social and Cultural Rights (CESCR) General Comment No 5 signifies that both the ICCPR and the ICESCR proscribe discrimination on the basis of disability.¹² General Comment No 5 is instructive and states that '*In order to remedy past and present discrimination, and to deter future discrimination, comprehensive anti- discrimination legislation in relation to disability would seem to be indispensable...*'¹³ General Comment No 5 also provides instruction on the definition of discrimination on the grounds of disability, which it states, '*any distinction, exclusion, restriction or preference, or denial of reasonable accommodation based on disability which has the effect of nullifying or impairing the recognition, enjoyment or exercise of economic, social or cultural rights.*'¹⁴ Interpreted in conjunction with General Comment No. 5, there appears to be wide scope in the ICCPR to protect against non- discrimination on a range of grounds including disability.

In a case that came before the Human Rights Committee (HRC), *Brough v Australia*,¹⁵ the rights of persons with disabilities was considered, in the context of children's rights (Article 2). The HRC found that the treatment of the individual in question was contrary to the ICCPR (Article 10 and Article 24) in light of the fact that he was a young person in a vulnerable situation, and on the grounds of his disability, and status as an Aboriginal.¹⁶ This case highlights the scope of the ICCPR. In further illustrating the scope of the ICCPR, it has been found that indirect discrimination is provided for under Article 26.¹⁷

The ICCPR also refers to a right to privacy for individuals, which is contained in Article 17, which provides that "*no one shall be subjected to arbitrary or unlawful interference with their privacy, family, home or correspondence, or to unlawful attacks on their honour or reputation.*"¹⁸ These provisions have been clarified by

¹⁰ ICCPR, Article 26

¹¹ Interights, *Non- Discrimination in International Law: A Handbook for Practitioners* (Interights: London, UK 2011) 195

¹² CESCR General Comment No. 5 (1994) Persons with Disabilities 12/09/1994

¹³ Ibid, para 16

¹⁴ Ibid, para 15

¹⁵ *Brough v Australia* (No. 1184/2003, ICCPR)

¹⁶ Interights, *Non- Discrimination in International Law: A Handbook for Practitioners* (Interights: London, UK 2011) 195

¹⁷ *Singh Bhinder v Canada* (No. 208/ 1986, ICCPR)

¹⁸ ICCPR, Article 17

General Comment No 16: Article 17.¹⁹ The ICCPR does not contain any specific references to genetics, but it does provide some guidance in terms of requiring free consent before medical or scientific “*experimentation*.”²⁰ In this regard, Article 7 states “...*no one shall be subjected without his free consent to medical or scientific experimentation*.”²¹ This article protects the autonomy and self-determination of individuals, particularly by indicating the need for informed consent as regards any “*experimentation*.” By implication, this Article also protects an individual’s privacy rights in this context.

2.2 International Covenant on Economic, Social and Cultural Rights

Against a backdrop of a fundamental human rights framework, the ICESCR focuses on the recognition and protection of economic, social and cultural rights.²² The recognition of these rights and an individual’s prerogative to enjoy these rights are core to recognising human dignity and integrity, and other fundamental rights. The enjoyment of these rights allows an individual to access further social goods and services, to interact with peers, and to fully participate in the community. A denial of these economic, social and cultural rights may have adverse implications for one’s dignity and integrity. It is submitted that such a denial of rights may have particularly undesirable implications for vulnerable groups such as persons with disabilities, the elderly, and those who are particularly susceptible to marginalisation in society.

The ICESCR also has a human rights focus and Article 1, mirrors Article 1 in ICCPR.²³ The ICESCR contains similar provisions to the ICCPR in Article 2, which provides that the ICESCR will be respected and ensured without distinction “... *of any kind such as race, colour, sex, language, religion, political or other opinion, national or social origin, property, birth or other status*.”²⁴ The right to work and earn a livelihood (for everyone) is a key feature of the ICESCR. For example, Article 6 sets out a general “*right of everyone to the opportunity to gain his living by work which he freely chooses or accepts*...” Article 7 sets out guidelines on the “...*right of everyone to the enjoyment of just and favourable conditions of work*...”²⁵ The inclusion of this right is particularly important as employment is effectively a gateway to accessing other social goods and services.

¹⁹ General Comment No. 16: Article 17 (Right to Privacy, Family, Home and Correspondence and Protection of Honour and Reputation) 8 April 1988, Adopted at 32nd Session of the Human Rights Committee

²⁰ Dinah Sheldon, ‘Challenges to the Future of Civil and Political Rights’ (1998) 55 Washington and Lee Law Review 3 669

²¹ ICCPR, Article 7

²² See generally Eibe Riedel, ‘Economic, Social and Cultural Rights’ in Catarina Krause and Martin Scheinin (eds) *International Protection of Human Rights: A Textbook* (2nd edn, Abo Akademi University Human Rights Institute 2012)

²³ ICESCR, Article 1

²⁴ ICESCR, Article 2

²⁵ ICESCR, Article 7

In addition, there are additional key rights granted in the ICESCR. For example, Article 12 provides for the right to “*the enjoyment of the highest attainable standard of physical and mental health*” and sets out conditions necessary to realising this right, which could potentially be interpreted to provide that all individuals should have access to genetic technologies, without fear of subsequent misuse.²⁶ Although disability is not specifically mentioned, the same rationale in respect of ‘other status’, as was argued in respect of the ICCPR, can be made here, and reference is made to General Comment No 5 for guidance in this respect. In acknowledging the benefits of science, Article 15 provides for the right of individuals to “*enjoy the benefits of scientific progress and its application.*” Article 15 further recognises the merits of fostering the development and progression of science.²⁷ This provision may indicate the need to ensure and enhance public confidence in the progression of genetic science and new technology.

It is noted that the ICESCR does not contain a similar provision to the ICCPR’s non- discrimination provision in Article 26. However, in this regard, an instructive case on Article 26 of the ICCPR is *Broeks v The Netherlands*,²⁸ in which the HRC held that it had the power to rule upon cases of discrimination as regards economic, social and cultural rights, as well as civil and political rights. Accordingly, it can be concluded that the HRC also has the jurisdiction to hear complaints relating to the economic, social and cultural rights of disabled persons.²⁹

2.3 Evaluation of the ICESCR and the ICCPR

On evaluation of the ICESCR and the ICCPR, it is evident that there is a clear human rights based approach towards recognising and guaranteeing a wide range of civil and political rights, as well as economic, social and cultural rights. The exercise and enjoyment of these rights are necessary to actively live in and contribute to the community. These treaties apply to all individuals, including persons with disabilities, as illustrated. The case law and the General Comments, discussed above, are particularly instructive on this issue. It is submitted that the application of these treaties to persons with disabilities is vital to ensure their full and meaningful participation in society. These treaties constitute a core element of international law and frame our understanding of fundamental human rights in this area. A common thread identified in both treaties is the reference to the

²⁶ ICESCR, Article 12

²⁷ ICESCR, Article 15 (1) and (4)

²⁸ *Broeks v The Netherlands* (No. 172/1984, ICCPR)

²⁹ Interights, *Non- Discrimination in International Law: A Handbook for Practitioners* (Interights: London, UK 2011) 196

'inherent dignity of the person.'³⁰ There is also a focus on equality and non-discrimination. As regards the application of these human rights to science and technology, Article 15 of the ICESCR is particularly relevant in implying the right to access scientific development for all individuals.

The treaties therefore provide a clear basis for understanding these fundamental human rights, and applying them to the current debate on genetic discrimination and protection of genetic privacy. In the context of this current debate, it is submitted that these rights are very much relevant and applicable. Our interpretation of human rights and civil rights evolve as societies evolve and as scientific, technological and other developments present new challenges, as well as legal and ethical dilemmas.³¹ One such challenge is that of advancing science and genetic technologies, which are becoming an increasing part of health care and indeed society.

3. United Nations Convention on the Rights of Persons with Disabilities

Against the backdrop of an evident gap in the protections and rights available to persons with disabilities, the UN introduced the CRPD and its Protocol in December 2006.³² The CRPD is the first international human rights treaty of the twenty-first century and its provisions have been described as providing "*a moral compass for change*,"³³ which aims to facilitate the fundamental human rights and equal treatment of persons with disabilities. As a holistic human rights treaty, it encompasses the broad range of life activities, including, employment, education, and health.³⁴ In addressing all facets of life, the CRPD therefore "*melds civil and political rights with economic, social and cultural rights*."³⁵ The CRPD affords disability rights a prime position within the framework of international human rights.³⁶ The principles of non-discrimination and equality feature strongly in the CRPD. Indeed, one of the main tools used in the CRPD is

³⁰ Nigel Rodley, 'Civil and Political Rights' in Catarina Krause and Martin Scheinin (eds) *International Protection of Human Rights: A Textbook* (2nd edn, Abo Akademi University Institute for Human Rights 2012) 106

³¹ Dinah Sheldon, 'Challenges to the Future of Civil and Political Rights' (1998) 55 Washington and Lee Law Review 3 669

³² CRPD

³³ Gerard Quinn, 'The United Nations Convention on the Rights of Persons with Disabilities: Toward a New International Politics of Disability' (2009- 2010) 15 Texas Journal on Civil Liberties and Civil Rights 1 33

³⁴ Michael Ashley Stein, 'A Quick Overview of the United Nations Convention on the Rights of Persons with Disabilities and Its Implications for Americans with Disabilities' (2007) 31 Mental and Physical Disability Law Reporter 679, 679

³⁵ Michael Ashley Stein and Janet Lord, 'Future Prospects for the United Nations Convention on the Rights of Persons with Disabilities' in Oddný Mjöll Arnardóttir and Gerard Quinn (eds) *UN Convention on the Rights of Persons with Disabilities* (Martinus Nijhoff Publishers 2009) 24

³⁶ Colm O'Cinneide, 'Extracting Protection for the Rights of Persons with Disabilities from Human Rights Frameworks: Established Limits and New Possibilities' in Oddný Mjöll Arnardóttir and Gerard Quinn (eds) *UN Convention on the Rights of Persons with Disabilities* (Martinus Nijhoff Publishers 2009) 163

that of non- discrimination. The CRPD emphasises the social model of disability, thereby focusing outward toward the environment and other external factors as creating barriers and disabling a person.

As explained, genetic testing can predict a low or a high susceptibility to or probability of future disability. Genetic discrimination can therefore be described as discrimination on the basis of putative disability, on the basis of genetic predisposition to disease, or on the basis of potential future disability.³⁷ Accordingly, if genetic information is too widely available, it exposes putative persons with disabilities to discrimination by interested third parties, therefore potentially violating the CRPD. As will be illustrated, the CRPD is drafted broadly with the objective of promoting inclusion of all individuals with disabilities, including those with putative disabilities and genetic predisposition to disability. In addition, the definition of disability discrimination and the objective of the substantive provisions of the CRPD can be interpreted as applying to the concept of genetic discrimination, thereby protecting genetic information. The CRPD also clearly speaks out in favour of maintaining privacy, which by implication, extends to genetic privacy. The next section will look at the relevant provisions of the CRPD.

3.1 The CRPD and genetic discrimination

Before examining the substantive provisions of the CRPD and the application to genetic discrimination, it is necessary to highlight the overall rationale and objective of the CRPD. The application of the CRPD to genetic discrimination is expressed primarily through the social model of disability, as well as the emphasis on the non- discrimination framework. This submission is further reinforced by the overall tenor and objectives of the CRPD, which focus on the core principles of human dignity and integrity. The next section will further explore the provisions of the CRPD through the lens of the social model of disability and the non- discrimination paradigm.

3.1.1 Social model of disability

The CRPD emphasises the paradigm shift away from the medical model of disability towards the social model of disability. As discussed in chapter 3, the social model views people with disabilities as rights holders and valued members of our societies who are often more disabled by the physical and attitudinal barriers societies erect to exclude and stigmatise them. In rejecting the medical model, the CRPD confronts outdated views of disability as an inherent medical

³⁷ See generally Paul S. Miller, 'Genetic Testing and the Future of Disability Insurance: Thinking about Discrimination in the Genetic Age' (2007) *Journal of Law, Medicine and Ethics* 47. See also Gregor Wolbring, 'A Disability Rights Approach to Genetic Discrimination' in Judit Sandor, (ed) *Society and Genetic Information: Codes and Laws in the Genetic Era* (Central European University Press 2004) 161

defect, giving rise to policies based on the principles of welfare, charity and pity.³⁸ In highlighting the CRPD's emphasis on the social model of disability, the Preamble (e) states that *"disability is an evolving concept and that disability results from the interaction between persons with impairments and attitudinal and environmental barriers that hinder their full and effective participation in society on an equal basis with others."*³⁹ It therefore challenges the physical and attitudinal barriers in society.

The CRPD has been described as *"the highest legal manifestation and confirmation of the social model of disability on the international stage."*⁴⁰ Indeed the rationale of many of the substantive articles of the CRPD, as well as the definitions reflect the ethos of the social model of disability. Article 1(2) also confirms the social model and states: *"Persons with disabilities include those who have long-term physical, mental, intellectual or sensory impairments which in interaction with various barriers may hinder their full and effective participation in society on an equal basis with others"*⁴¹ reaffirming the theory that limitations arise as a result of the interaction with the various barriers in society.⁴² It has been observed that this is not necessarily a definition, but more accurately, a bare minimum, and therefore, any interpretation of the CRPD should include not only persons with disabilities, but also persons who have genetic predispositions to disability, and those to whom disabilities are imputed.⁴³

Article 3 sets out the guiding principles in the Convention. The main values or principles that inform the CRPD include dignity, autonomy, non- discrimination, full and active participation and inclusion, respect for difference and human diversity, and equality of opportunity.⁴⁴ These principles are reflective of the paradigm shift to the social model of disability and are intended to inform the

³⁸ Janet E. Lord and Rebecca Brown, 'The Role of Reasonable Accommodation in Securing Substantive Equality for Persons with Disabilities: The UN Convention on the Rights of Persons with Disabilities' in Marcia H. Rioux, Lee Ann Bassar and Melinda Jones (eds) *Critical Perspectives on Human Rights and Disability Law* (Martinus Nijhoff Publishers 2011). Available on SSRN: http://papers.ssrn.com/sol3/papers.cfm?abstract_id=1618903 (accessed 26 August 2013) at p 24

³⁹ Delia Ferri, 'The Conclusion of the UN Convention on the Rights of Persons with Disabilities by the EC/EU' in Gerard Quinn and Lisa Waddington (eds) *2 European Yearbook of Disability Law* (Intersentia 2010) 52

⁴⁰ Lisa Waddington, 'Breaking New Ground: The Implications of Ratification of the UN Convention on the Rights of Persons with Disabilities for the European Community' in Oddný Mjöll Arnardóttir and Gerard Quinn (eds) *UN Convention on the Rights of Persons with Disabilities* (Martinus Nijhoff Publishers 2009) 115

⁴¹ CRPD, Article 1

⁴² Michael Ashley Stein and Janet Lord, 'Future Prospects for the United Nations Convention on the Rights of Persons with Disabilities' in Oddný Mjöll Arnardóttir and Gerard Quinn (eds) *UN Convention on the Rights of Persons with Disabilities* (Martinus Nijhoff Publishers 2009) 25

⁴³ Delia Ferri, 'Legislating at EU level: Challenges and Possibilities' at 'Genetic Discrimination - Transatlantic Perspectives on the Case for a European Level Legal Response' (19 November 2011, Galway, Ireland)

⁴⁴ CRPD, Article 3

CRPD's interpretation.⁴⁵ These values are important in terms of acting as a useful guidance mechanism, and a tool to interpret and clarify some of the ambiguities that may be present in the Convention.⁴⁶

It is noted that discrimination on the basis of genetic information can result in a violation of the above principles, resulting particularly in denial of equal opportunities and eventually isolation from the community. It is submitted that the existence of barriers to employment, insurance and other social goods (as a consequence of misusing genetic information) may arguably result in the exclusion of certain vulnerable individuals and may violate the core principles of dignity and integrity. In other words, it is acknowledged that employment and insurance are portals to accessing other social and economic goods and services, which can impact upon an individual's participation in society. In addition, the reference to 'difference' and 'human diversity' acknowledge the intrinsic differences between individuals and the need to respect genetic differences between individuals.

3.1.2 A non- discrimination framework

In terms of a legal framework, *"the convention seems to let the non-discrimination tool do most of the heavy lifting,"*⁴⁷ and in this regard, the principles of equality and non- discrimination are key underlying concepts. Through this framework, and in line with the social model of disability, the CRPD facilitates higher standards of non- discrimination and equality with respect to persons with disabilities. Non- discrimination as a key premise in the CRPD is referred to in the Preamble (h), which states *"...discrimination against any person on the basis of disability is a violation of the inherent dignity and worth of the human person."*⁴⁸ This formulation reflects the core ethos of the non- discrimination and equality principles. The non- discrimination principle is also reflected in Article 3(b) on the general principles. The CRPD adopts a clear non- discrimination framework against the backdrop of a social model approach to disability.

3.1.3 Definition of discrimination on the basis of disability

Before looking at the substantive articles of the CRPD, this section will analyse

⁴⁵ Anna Lawson, 'The UN Convention on the Rights of Persons with Disabilities and European Disability Law: A Catalyst for Cohesion?' in Oddný Mjöll Arnardóttir and Gerard Quinn (eds) *UN Convention on the Rights of Persons with Disabilities* (Martinus Nijhoff Publishers 2009) 88

⁴⁶ Gerard Quinn, 'Disability and Human Rights: A New Field in the United Nations', in Catarina Krause and Martin Schenin (eds) *International Protection of Human Rights: A Textbook* (Gummerus Printing 2009)

⁴⁷ Gerard Quinn, 'Disability and Human Rights: A New Field in the United Nations' in Catarina Krause and Martin Schenin (eds) *International Protection of Human Rights: A Textbook* (Gummerus Printing 2009) 257

⁴⁸ Preamble (h)

the definition of discrimination on the basis of disability. In line with the holistic and inclusive nature of the CRPD, there is a broad approach taken to the definition of discrimination on the basis of disability. It is submitted that discrimination on the basis of genetic information, or on the basis of genetic predisposition to disability constitutes discrimination on the grounds of disability, as provided for in the CRPD.

Pursuant to Article 2,⁴⁹ discrimination on the basis of disability as anticipated by the CRPD therefore includes all forms of discrimination; direct, indirect, structural, multiple or other, as well as discrimination by association and discrimination based on assumed or future disability.⁵⁰ The wording of ‘on the basis of disability’ is also relevant in taking the focus away from the individual and placing it on the actions and attitudes of third parties, again reflecting the social construction of disability. Consequently, discrimination on the basis of genetic information could very well come within the ambit of this definition. The next section will look at some of the substantive provisions of the CRPD.

3.2 Relevant provisions of the CRPD

3.2.1 Article 5

In reaffirming the main objectives of the CRPD, Article 5 provides specifically for the principles of equality and non- discrimination. It requires, *inter alia*, the prohibition of discrimination ‘on the basis of disability’ and guarantees to persons with disabilities equal and effective legal protection against discrimination on all grounds.⁵¹ It has been observed that the term discrimination ‘on the basis of disability’ is “*wider than a definition that focuses on the peculiar impairments of any given individual.*”⁵² Discrimination on the basis of disability as provided for in the CRPD has the potential to encompass individuals who are not themselves disabled but who are discriminated against on the grounds of their “*association with an individual who has a disability.*”⁵³ As noted, the definition of discrimination takes the focus away from the individual and places it on the actions of third parties.

Article 5, with its focus on the actions of third parties and read in conjunction with the overall intention of the CRPD could arguably be interpreted as applying to discrimination on the basis of genetic information or genetic predisposition to disability. Article 5 provides a core basis for equality and non- discrimination and

⁴⁹ CRPD, Article 2

⁵⁰ European Foundation Centre, Study on challenges and good practices in the implementation of the UN Convention on the Rights of Persons with Disabilities VC/ 2008/ 1214 Final Report, p 54

⁵¹ CRPD, Article 5

⁵² Gerard Quinn, ‘Disability and Human Rights: A New Field in the United Nations’ in Catarina Krause and Martin Schenin (eds) *International Protection of Human Rights: A Textbook* (2nd ed, Abo Akademi University for Human Rights 2012) 279

⁵³ *Ibid* at 279

highlights the main approach taken towards ensuring the rights of persons with disabilities.

3.2.2 Article 27

In continuing the theme of non- discrimination, Article 27 focuses on work and employment, recognising the right of persons with disabilities to work on an equal basis with others, and prohibiting discrimination on the basis of disability.⁵⁴ Article 27 (a) “*prohibits discrimination on the basis of disability with regard to all matters concerning all forms of employment, including conditions of recruitment, hiring and employment, continuance of employment, career advancement...*” An expansive interpretation of Article 27 would prohibit discrimination on the grounds of genetic information during all the stages of employment listed in Article 27 (a), including during the recruitment stage and throughout employment, for example in relation to promotional opportunities. As discussed in chapter 3, employers generally want to hire the most healthy and productive employees, and those who will be the least draining on an employer’s resources. Use of an employee’s genetic information may result in discrimination and the denial of employment opportunities on the basis of perceived disability. Such practices may therefore create unfair barriers to employment that may exclude certain individuals from the labour market.

In conjunction with the overall objectives and rationale of the CRPD, particularly the principles of equality and non- discrimination, Article 27 could therefore be interpreted to cover discrimination on the basis of genetic information in employment. Such an interpretation is also in line with underlying principles of inclusion and active participation in society, which the CRPD provides for. By creating barriers to employment, this may result in the unjustified exclusion of certain vulnerable individuals, with undesirable social consequences, as explained in chapter 3. Such access to employment is also necessary in ensuring an individual’s rights to dignity and integrity. It is noted that the scope and coverage of the Convention is unparalleled.⁵⁵ An expansive interpretation of Article 27 to include discrimination on the basis of genetic predisposition to disability would therefore seem appropriate and in line with the overall objectives of the CRPD.

An in-depth analysis of Article 27 may strike a balance between the competing rights between employers and employees, and recognise the potentially legitimate interests of employers in certain circumstances. Article 27 places an emphasis on employers’ obligations as regards ‘safe and healthy working conditions.’⁵⁶ As regards genetic testing, this provision might be interpreted as

⁵⁴ CRPD, Article 27

⁵⁵ Arlene Kanter, ‘The Promise and Challenge of the United Nations Convention on the Rights of Persons with Disabilities’ (2006- 2007) 34 Syracuse Journal of International Law and Commerce 287, 289

⁵⁶ CRPD, Article 27 (1)(b)

giving rise to a limited right on the part of an employer to engage in genetic testing to address occupational disease and in response to health and safety obligations, under Article 27 and under existing EU health and safety legislation.⁵⁷ This would acknowledge an individual's right to equality in employment, and the corresponding limited right of an employer to use genetic information, under strict conditions.

3.2.3 Article 25

Article 25 is also relevant in further highlighting the non-discrimination framework.⁵⁸ Article 25 provides that persons with disabilities are entitled to the highest standard of health without discrimination 'on the basis of disability.' The freedom to undergo genetic testing and to take advantage of advancing genetic technologies may indeed contribute towards enhancing the health of all individuals, and may operate to minimise and prevent further potential disabilities. Of particular interest is Article 25(b) which stipulates that the provision of additional services is needed because of disability (including early identification, intervention as appropriate), to "*minimize and prevent further disabilities*."⁵⁹

Further, Article 25(e) prohibits discrimination against persons with disabilities in the provision of health insurance, and life insurance where such insurance is permitted by national law. The refusal of health or life insurance in these circumstances or the imposition of prohibitively high insurance premiums on the basis of genetic information or genetic test results, may operate to push certain individuals out of the insurance pools, thereby creating barriers to health care and other services. As noted, genetic technologies are becoming increasingly available to individuals, offering further potential knowledge of disease and disability, and the possibility to effectively manage health care. However, there is a potential for misuse of such information, such as discrimination. The fear of this misuse may result in some individuals being reluctant to engage in genetic testing that may benefit their health, and therefore may act as a barrier to access to health care. In order to eliminate this fear, and instil confidence in individuals, appropriate regulation is necessary, allowing access to medical advances for all individuals.

3.2.4 Article 22

⁵⁷ Employers have obligations under European health and safety law, and under common law: see chapter 3

⁵⁸ CRPD, Article 25

⁵⁹ Gerard Quinn and Charles O'Mahony, 'Disability and Human Rights' in Catarina Krause and Martin Scheinin (eds) *International Protection of Human Rights: A Textbook* (2nd edn, Abo Akademi University Institute for Human Rights 2012) 295

Article 22 in relation to respect for privacy merits discussion.⁶⁰ Article 22(1) states “No person with disabilities... shall be subjected to arbitrary or unlawful interference with his or her privacy, family, home or correspondence or other types of communication or to unlawful attacks on his or her honour and reputation...”⁶¹ Access to and disclosure of genetic information to third parties without consent interferes with the right to privacy. It is arguable that data protection laws address the issue of genetic privacy by regulating the access to and disclosure of information.⁶²

3.3 Other relevant provisions

There are a number of other provisions of the CRPD that are relevant to this discussion. Article 17 protects the integrity of the person and provides that every person with disabilities has a right to respect for his or her physical and mental integrity on an equal basis with others. Genetic science could operate as a tool of social manipulation and ultimately lead to the practice of genetic cleansing.⁶³ If this practice takes hold, and a societal perception towards “*genetic perfection*” develops, this may result in further negative attitudes towards persons with disabilities as being inferior.⁶⁴ This may also have undesirable societal implications by categorising individuals according to what may be deemed to be genetic status.

Article 8 on awareness raising is also relevant, particularly in terms of attitudes to disability and attitudes to and perceptions of genetic predispositions to disability. Negative attitudes towards disability and genetic conditions may result in stigmatisation and exclusion. Fostering environments of positive attitudes towards disability can help to improve attitudes towards genetic conditions, as well as attitudes towards genetic predispositions. Article 8 requires a publicity campaign to ‘nurture receptiveness to the rights of persons with disabilities’ and to promote recognition of the ‘skills, merits and abilities of persons with disabilities.’⁶⁵ It also requires states to encourage all organs of the media to ‘portray persons with disabilities in a manner consistent with the purpose of the’ Convention.⁶⁶ Interpretation of this article might therefore manifest in the form of educational and awareness raising campaigns.

⁶⁰ CRPD, Article 22

⁶¹ CRPD, Article 22 (1)

⁶² For further discussion see chapter 9

⁶³ Iulia V. Motoc, ‘The International Law of Genetic Discrimination: The Power of Never Again’ in Therese Murphy (ed) *New Technologies and Human Rights* (Oxford University Press 2009) 243

⁶⁴ Ibid at 242

⁶⁵ CRPD, Article 8

⁶⁶ Ibid

In this regard, the introduction of the CRPD may represent an opportunity to tackle attitudinal barriers toward disability, in the form of awareness raising.⁶⁷ It is submitted that any such awareness raising take a broad approach and encompass attitudes towards genetic predispositions and conditions. Quinn has also referred to the importance of this provision since “*a culture shift in attitudes and value could make the difference between rhetorical change and real improvement.*”⁶⁸

Finally, reference is made to Article 33, which provides for the setting up of mechanisms with a view to monitoring the implementation of the CRPD. This is an innovation of the international human rights framework.⁶⁹

3.3.1 Evaluation of the substantive provisions of the CRPD

On examining the substantive provisions of the CRPD, it is clear that there is a focus on the framework of non- discrimination, as a means to achieve fulfilment of the rights of persons with disabilities. This approach is complemented by a corresponding focus on the notion of human dignity and integrity. There is an emphasis on the social model of disability as a central paradigm. It is asserted that the substantive articles of the CRPD can be interpreted to encompass the protection of genetic information – for example, by preventing discrimination on the basis of such information, protecting privacy, facilitating access to health care and awareness raising to promote positive attitudes towards disability and genetic predispositions to disability. The CRPD provides a clear international platform to address these issues.

In highlighting the potential application of the CRPD to the area of genetic discrimination and the concept of imputed disability, the case of *Kenneth McAlpine v United Kingdom* is of relevance.⁷⁰ On foot of the Optional Protocol, this claim was brought before the CRPD Committee and was the second individual petition to the Committee under the Optional Protocol. This case against the United Kingdom involved an individual with diabetes who was made redundant from his employment. He claimed that his redundancy was based on the assumption that diabetes results in prolonged periods of absence due to illness, and consequently that he had been the victim of disability based discrimination. The claim was therefore based on the notion of stereotypes and imputed disability. Although the Committee found that the complaint was inadmissible, the relevance of this case lies particularly in the fact that the particular area concerning imputed disability was one of the first complaints

⁶⁷ Elizabeth Emens, ‘Disabling Attitudes: U.S. Disability Law and the ADA Amendments Act’ (2012) 60 American Journal of Comparative Law 205

⁶⁸ Gerard Quinn, ‘Disability and Human Rights: A New Field in the United Nations’ in Catarina Krause and Martin Schenin (eds) *International Protection of Human Rights: A Textbook* (Gummerus Printing 2009) 261

⁶⁹ CRPD, Article 33

⁷⁰ *Kenneth McAlpine v United Kingdom* (No. 6/ 2011 UN CRPD)

brought using the Optional Protocol.⁷¹

3.4 The conclusion of the CRPD by the EU

This section will highlight the actions of the EU in concluding the CRPD, and the potential implications this may have on the EU's responsibilities and obligations in this area. The EU concluded the CRPD on December 2010, and it came into force on 22 January 2011.⁷² This historic action represents the first time that the EU has concluded an international human rights treaty. The EU has not concluded the UN treaties discussed in part one of this chapter. The EU's legal basis to sign the CRPD derived from Article 13 EC (now Article 19 TFEU) addressing (disability) discrimination and Article 95 EC (now Article 114 TFEU) addressing the internal market.

The conclusion of the CRPD by the EU represents a firm commitment to human rights, and the protection of disability rights. In this regard, it has been observed that the CRPD "*will take on a completely new legal status within EC law, and will have to be respected in its totality, and not as a mere add on to be referred to in passing in Community instruments,*"⁷³ expressing the view that this step by the EU signals positive action. However, although this development is a welcomed promotion of human rights and recognition of disability rights, it raises many challenging questions for the EU.⁷⁴ In particular, questions arise as regards the particular obligations and legislative competence of the EU in this area, which will be discussed in the following section.

3.4.1 EU obligations under the CRPD

International treaties have long been regarded as forming "*an integral part of Community law*"⁷⁵ once they entered into force. The submission that international law forms an integral part of EU law appears to be upheld by cases such as *Intertanko* and *Kadi*.⁷⁶ As regards the status of international agreements, it has

⁷¹ Ibid

⁷² Council Decision 2010/48/EC of 26 November 2009 concerning the conclusion, by the European Community of the United Nations Convention on the Rights of Persons with Disabilities [2010] OJ L 23/35

⁷³ Lisa Waddington, 'A New Era in Human Rights Protection in the European Community: The Implications the United Nations Convention on the Rights of Persons with Disabilities for the European Community' (2007) 4 Maastricht Faculty of Law Working Paper, 2. See also Grainne de Burca, 'The EU in the negotiation of the UN Disability Convention' (2010) 35 European Law Review 2

⁷⁴ Lisa Waddington, 'The European Union and the United Nations Convention on the Rights of Persons with Disabilities: A Story of Exclusive and Shared Competences' (2011) 18 Maastricht Journal 4

⁷⁵ Case C- 181/73 R. & V. *Haegeman v Belgian State* [1974] ECR 449, para 5

⁷⁶ Joined cases C-402/05 P and C-415/05 P *Yassin Abdullah Kadi and Al Barakaat International Foundation v. Council of the European Union and Commission of the European Communities* [2008] ECR I-6351. See also Ramses Wessel, 'Reconsidering the Relationship between International and EU

been confirmed that international treaties are situated formally below the provisions of the Treaties.⁷⁷

In ascertaining the EU's potential obligations as a result of concluding the CRPD, it is necessary to outline the nature of the CRPD as a legal document, as well as the competence of the EU to act in this area. The CRPD is an example of a "*mixed agreement*", meaning that part of an international agreement is within the scope of the powers and responsibilities of the EU, and part of an agreement is within the scope of the powers and responsibilities of the Member States.⁷⁸ Tackling discrimination on the grounds of disability is an area of shared competence between the EU and the Member States.⁷⁹ It has been asserted that it is unlikely that there is a legal obligation on the EU under the CRPD to take proactive measures.⁸⁰ It follows that any EU action that is taken must be in compliance with the CRPD, but there does not seem to be any legal obligation to act in the fields of shared competence. Therefore, the EU, as a State Party is not legally obliged to act in this area. Waddington however, expresses the view that it can nevertheless be "*desirable for the EU to act in such areas of shared competence*."⁸¹

Therefore, in terms of fulfilling its obligations under the CRPD, the EU could adopt a conservative approach, ensuring that the EU must simply not breach the provisions of the CRPD when it acts in areas of shared competence. Alternatively, the EU could take a more expansive approach, particularly in light of the fact that this is the first instance of the EU concluding an international human rights treaty. There are several arguments that can be made in support of this submission. One of the arguments in favour of EU action in areas of shared competence is to prevent the risk of differing national CRPD implementation, resulting in diverging standards and levels of protection throughout the EU. This may have a negative impact on the internal market in creating barriers. Concerted action by the EU is therefore the most practical and efficient means of effectively implementing the CRPD. Such uniform EU legislative action can help to guard against the introduction of varying national approaches and can facilitate the effective implementation of the CRPD as well as other goals, such as

Law: Towards a Content-Based Approach?', in Enzo Cannizzaro, Paolo Palchetti, Ramses A. Wessel (eds) *International Law as Law of the European Union* (Martinus Nijhoff Publishers 2011)

⁷⁷ Case C-61/94 *Commission v. Germany* [1996] ECR I-3989, para 52

⁷⁸ Delano Verwey, 'The European Community, the European Union and the International Law of Treaties: A Comparative Legal Analysis of the Community and the Union's External Treaty- Making Practice' (T.M.C. Asser Press 2004) 35

⁷⁹ Delia Ferri, 'The Conclusion of the UN Convention on the Rights of Persons with Disabilities by EC/EU: Some Reflections from a "Constitutional" Perspective', Centro di documentazione europea – Università di Catania – Online Working Paper 2010 – Serie Speciale/ n.4 (March 2010). See also Case C- 239/03 *Etang de Berre* [2004] ECR I- 9325, para 25

⁸⁰ Lisa Waddington, 'The European Union and the United Nations Convention on the Rights of Persons with Disabilities: A Story of Exclusive and Shared Competences' (2011) 18 *Maastricht Journal* 4 431, 448

⁸¹ *Ibid* at 448

ensuring the operation of the EU internal market.⁸²

There are of course opposing arguments to the effect that national legal systems and traditions of the individual Member States would be adversely impacted by EU action. However, in trying to reach a balance, it is submitted that potential EU legislative action (in the form of a directive) would represent a compromise between fulfilling the EU's obligations in implementing the CRPD and achieving comprehensive EU level protection, while at the same time leaving scope in implementation for national approaches.

Pursuant to the CRPD, the EU is under an obligation not only to respect human rights but also to protect and fulfill them.⁸³ These provisions would seem to indicate a preference for a positive, proactive approach by the EU in this area. In addition, the desire for EU consistency and uniformity in standards would also point towards EU level action in terms of implementing the Convention.⁸⁴

On looking at the EU conclusion of the CRPD and the obligations that ensue, Article 4 is important. Article 4 sets out the general obligations and the governing principles by which States Parties are obliged to act. These general obligations include an undertaking to adopt new legislation and other appropriate measures where needed to implement the Convention, to modify or repeal laws, customs or practices that constitute disability discrimination, to mainstream disability into all relevant policies and programmes, to refrain from any act or practice that is inconsistent with the convention, to take all appropriate measures to eliminate discrimination on the basis of disability by any person, organisation or private enterprise.⁸⁵

The CRPD therefore imposes certain obligations to enact legislation, particularly with a view to eliminating discrimination on the basis of disability. It was not intended as a mere guidance document. On reflecting upon the merits of this provision, Quinn has stated that this article "*converts the convention into a trigger for worldwide disability law reform.*"⁸⁶ In light of the fact that the EU has concluded the CRPD, the question arises whether this article could trigger disability reform in the area of genetic discrimination in the EU. In this regard, any potential legislation in this field could be regarded as implementation of the Convention, with a view to eliminating discrimination on the basis of genetic predisposition to disability. Therefore, the objective of Article 4 would seem to support positive EU level action in this area to implement the CRPD.

⁸² Ibid at 451

⁸³ UN CRPD, Article 1

⁸⁴ Lisa Waddington, 'The European Union and the United Nations Convention on the Rights of Persons with Disabilities: A Story of Exclusive and Shared Competences' (2011) 18 Maastricht Journal 4 431, 453

⁸⁵ CRPD, Article 4

⁸⁶ Gerard Quinn, 'The United Nations Convention on the Rights of Persons with Disabilities: Towards a New International Politics of Disability' (2009) 15 Texas Journal on Civil Liberties and Civil Rights 1 33, 41

On looking at the impact of the CRPD on the EU legal order, it is noted that the ECJ has referred to the CRPD in its interpretation of the Employment Equality Directive.⁸⁷ Similarly the ECtHR has also referred to the CRPD in its case law.⁸⁸

3.5 The CRPD as a catalyst for change

The CRPD is in a key position to initiate reform and to provoke “*a more comprehensive and more cohesive European disability equality law.*”⁸⁹ The CRPD could well act as a catalyst to develop a wave of reform around the regulation of genetic information and develop a deeper thinking around the concept of disability and disability discrimination, to encompass potential future disability, putative disability and genetic predisposition to disability. In this context, it has been opined that the CRPD “*seems likely to put fresh pressure on the EU to broaden and intensify reform in the field of disability discrimination.*”⁹⁰ In line with the paradigm shift to acceptance of the social model, it is submitted that such a forward- thinking approach be taken to the issue of genetic information non- discrimination. Since its introduction, the Convention has generated a significant momentum in terms of instigating reform.⁹¹ Perhaps, this momentum can continue in addressing the regulation of genetic information.

In conjunction with this submission, the CRPD has been described as “*an expressive value trigger.*”⁹² The area of expressive law looks at the potential of law to influence behavioural patterns and shape societal outlooks and values.⁹³ In the context of considering the regulation of genetic information, the CRPD, as an “*expressive value trigger,*” can engrain a message and drive a movement acknowledging the value of protecting genetic information while at the same time allowing science and technology to flourish. The CRPD has the potential to provoke law reform and act as an impetus for EU level action, as well as providing a clear rights- based framework from which to address these issues.

⁸⁷ Joined cases C- 335/11 and C- 337/11 *HK Danmark, acting on behalf of Jette Ring v Dansk almennyttigt Boligselskab* (C-335/11) and *HK Danmark, acting on behalf of Lone Skouboe Werge v Dansk Arbejdsgiverforening, acting on behalf of Pro Display A/S, in liquidation* (C-337/11). See also Case C-312/11 *European Commission v Italy* [2013] (4 July 2013)

⁸⁸ *Glor v Switzerland* (No. 13444/04) [2009] ECHR 13 April 2009

⁸⁹ Anna Lawson, “The UN Convention on the Rights of Persons with Disabilities and European Disability Law: A Catalyst for Cohesion?” in Oddný Mjöll Arnardóttir and Gerard Quinn (eds) *UN Convention on the Rights of Persons with Disabilities* (Martinus Nijhoff Publishers 2009) 109

⁹⁰ Grainne de Burca, “The Trajectories of European and American Antidiscrimination Law” (2012) 60 *American Journal of Comparative Law* 1 12

⁹¹ *Ibid* at 109

⁹² Michael Ashley Stein and Janet Lord, ‘Future Prospects for the United Nations Convention on the Rights of Persons with Disabilities’ in Oddný Mjöll Arnardóttir and Gerard Quinn (eds) *UN Convention on the Rights of Persons with Disabilities* (Martinus Nijhoff Publishers 2009) 31- 32

⁹³ *Ibid*. See also Robert Cooter, ‘Expressive Law and Economics’ (1998) 27 *Journal of Legal Studies* 585; Richard H. McAdams, ‘An Attitudinal Theory of Expressive Law’ (2000) 79 *Oregon Law Review* 339

4. Relevant International Soft Law: The United Nations Educational, Scientific and Cultural Organisation Declarations

Having examined the UN treaty law and its impact in shaping human rights in this area, this section will highlight the UN soft law instruments and their potential to inform our understanding of this area. As a general observation, it is submitted that the overall tenor of the international instruments is one that reflects a past history of abuse of science and eugenic tendencies.⁹⁴ Following the eugenics disasters which occurred during World War II, international law sought to regulate and outlaw discrimination on the basis of genetics, in the knowledge that such discrimination can potentially lead to eugenic type practices.⁹⁵ In support of the merits of soft- law instruments, it has been observed that “*if the same non-binding standards are reaffirmed in successive declarations, in the course of time they may become binding rules... as it happened with the Universal Declaration of Human Rights of 1948.*”⁹⁶ The advantages of such a soft law approach in this area have been recognised by UNESCO.

It is further pointed out that the approach of UNESCO is reflective of the concept of genetic exceptionalism,⁹⁷ recognising the need to single out genetic information as being worthy of special protection (as discussed in chapter 5). The approach is also reflective of core values of non- discrimination and equality as guiding principles. Human dignity is also a common thread running through the UNESCO Declarations in this area.⁹⁸ The concept of human dignity is core to respect for genetic difference, and acknowledging the unique nature of genetic information. In addition, the reliance on ‘human dignity’ also reflects the submission that the protection of genetic information is a core human rights issue.⁹⁹ The following section will explore the three primary UNESCO Declarations.

4.1 The Universal Declaration on the Human Genome and Human Rights

⁹⁴ Iulia V. Motoc, ‘The International Law of Genetic Discrimination: The Power of Never Again’ in Therese Murphy (ed) *New Technologies and Human Rights* (Oxford University Press 2009) 225

⁹⁵ Ibid

⁹⁶ Roberto Andorno, ‘Global Bioethics at UNESCO: in defence of the Universal Declaration on Bioethics and Human Rights’ (2007) 33 *Journal of Medical Ethics* 150, 151

⁹⁷ Sirpa Soina, ‘Genetic Testing legislation in Western Europe – a fluctuating regulatory target’ (2012) 3 *Journal of Community Genetics* 143, 145

⁹⁸ Souheil El- Zein, ‘Genetic Manipulation: How to Strike the Right Balance between Technology and Respect for Human Rights’ in Antonio Cassese (ed) *Realizing Utopia- The Future of International Law* (Oxford University Press 2012) 489

⁹⁹ Jennifer Elle Tauer, ‘International Protection of Genetic Information: The Progression of the Human Genome Project and the Current Framework of Human Rights Doctrines’ (2000- 2001) 29 *Denver Journal of International Law and Policy* 209, 235

In 1997, UNESCO adopted the Universal Declaration on the Human Genome and Human Rights.¹⁰⁰ It was the first universal instrument to introduce an ethical structure for activities in the area of bioethics and the human genome, which placed the concept of human dignity at the forefront, along with other human rights.¹⁰¹

The relevance of the Preamble is noted, which states that “*the recognition of the genetic diversity of humanity must not give rise to any interpretation of a social or political nature which could call into question ‘the inherent dignity and... the equal and inalienable rights of all members of the human family’....*”¹⁰² This Declaration therefore highlights the notion that the unique genetic profile of an individual is not definitive of the inherent value and worth of an individual.¹⁰³ In particular, the Preamble denounces the use of science to promote ideas or objectives that are opposed to equality and freedom,¹⁰⁴ thereby implying the condemnation of policies that use genetics as a means to express undesirable social behaviour and prejudice.¹⁰⁵

The main theme of the 1997 Declaration is the protection of human dignity, as reflected for example in Article 1.¹⁰⁶ In continuing the theme of respect for human dignity and implying the prohibition of genetic discrimination, Article 2 states, “(a) *Everyone has the right to respect for their dignity and for their human rights regardless of their genetic characteristics. (b) That dignity makes it imperative not to reduce individuals to their genetic characteristics and to respect their uniqueness and diversity.*”¹⁰⁷ This provision also implies disapproval of the concept of genetic determinism and the practice of genetic cleansing. From an ethical perspective, it is observed that an individual should not be reduced to the sum of their genes (as discussed in chapter 3).

In indicating support of a non- discrimination framework, Article 6 prohibits “*discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights...*”¹⁰⁸ These non- discrimination provisions are also reflected in the Preamble. This Declaration further states the importance of maintaining the confidentiality of genetic data,¹⁰⁹ the need for informed

¹⁰⁰ G.A. Res. 152, UN GAOR, 53rd Sess., UN. Doc A/53/625/Add.2 (1998) [“UDHGHR”]

¹⁰¹ Deryk Beyleveld and Roger Brownsword, ‘Human Dignity, Human Rights, and Human Genetics’, (1998) 61 Modern Law Review 658, 664

¹⁰² UDHGHR, Preamble

¹⁰³ Noelle Lenoir, ‘Universal Declaration on the Human Genome and Human Rights: The First Legal and Ethical Framework at the Global Level’ (1998- 1999) 30 Columbia Human Rights Law Review 537, 561

¹⁰⁴ UDHGHR, Preamble

¹⁰⁵ Noelle Lenoir, ‘Universal Declaration on the Human Genome and Human Rights: The First Legal and Ethical Framework at the Global Level’ (1998- 1999) 30 Columbia Human Rights Law Review 537, 561

¹⁰⁶ UDHGHR, Article 1

¹⁰⁷ UDHGHR, Article 2

¹⁰⁸ UDHGHR, Article 6

¹⁰⁹ UDHGHR, Article 7

consent and the right of individuals to decide whether they want to be informed of a genetic condition.¹¹⁰ The Declaration also recognises the merits of genetic science and acknowledges the need to ensure that science and research advance for the benefit of all. This is reflected in Article 17, which provides, *inter alia*, States ... *should foster, inter alia, research on the identification, prevention and treatment of genetically based and genetically influenced diseases...*¹¹¹ It is interesting to note that Article 4 implies a rejection of property rights in genetic information, and states “*The human genome in its natural state shall not give rise to financial gain.*”¹¹²

This Declaration therefore speaks strongly in favour of protecting genetic diversity and prohibiting discrimination, while at the same time recognising the great potential of genetic science for improving the health of individuals. Although the Declaration does not reflect a distinct regulatory approach, the Declaration certainly advocates the concept of non- discrimination in this area, with a clear focus on maintaining human dignity. There is also an acknowledgement to respect privacy and confidentiality when dealing with genetic information. The Declaration was an important first step in international law in setting ethical and moral standards in response to genetic discoveries.¹¹³ It therefore has a strong expressive and symbolic value in this area. It also highlights the evolution of human rights, and the need to interpret human rights in light of changing societal norms and other scientific and technological changes.

4.2 The International Declaration on Human Genetic Data

In 2003, UNESCO adopted the International Declaration on Human Genetic Data,¹¹⁴ which has been described as “*a sequel*” to the Universal Declaration on the Human Genome and Human Rights.¹¹⁵ The 2003 Declaration conveys similar objectives to the 1997 Declaration; specifically to “*ensure the respect of human dignity and protection of human rights and fundamental freedoms in the collection, processing, use and storage of human genetic data...*”¹¹⁶ In addition, this Declaration also contains similar provisions denouncing the notion of genetic determinism and provides that “*a person’s identity should not be reduced to genetic characteristics...*”¹¹⁷ However, the provisions in the 2003 Declaration are

¹¹⁰ UDHGHR, Article 5

¹¹¹ UDHGHR, Article 17

¹¹² UDHGHR, Article 4

¹¹³ Jennifer Elle Tauer, ‘International Protection of Genetic Information: The Progression of the Human Genome Project and the Current Framework of Human Rights Doctrines’ (2000- 2001) 29 Denver Journal of International Law and Policy 209, 231- 237

¹¹⁴ International Declaration on Human Genetic Data adopted by UNESCO’s General Conference on 16 October 2003 [“IDHGD”]

¹¹⁵ Abdulkawi A. Yusuf, ‘UNESCO Standard- setting Activities on Bioethics: Speak Softly and Carry a Big Stick’ in Francesco Francioni (ed) *Biotechnologies and International Human Rights* (Hart Publishing 2007) 85, 91

¹¹⁶ IDHGD, Article 1

¹¹⁷ IDHGD, Article 3

more specific than those in the 1997 Declaration and it reflects the reality of advancing genetic technology, while at the same time recognising the legal and ethical issues that arise.

This instrument establishes guidelines that should govern the collection, processing, use and storage of human genetic data. Privacy and confidentiality are specifically provided for in Article 14.¹¹⁸ Although the Declaration focuses on the importance of privacy protections, it also includes a provision on non-discrimination and non-stigmatization.¹¹⁹ As noted, UNESCO takes the approach of genetic exceptionalism. This is reflected particularly in Article 4 of the 2003 Declaration, which affords particular importance to human genetic data because such information can be predictive of genetic predisposition; the information impacts not only the individual, but also family members; and it may contain information that is not yet known.¹²⁰

This Declaration is important in promoting acknowledgement of advancing genetic science and the corresponding ethical considerations that arise. In mirroring the 1997 Declaration, this Declaration advocates primarily a non-discrimination framework. In terms of providing guidance on an appropriate regulatory framework, it also places emphasis on the key principles of privacy and human dignity. It builds upon the 1997 Declaration and further highlights the need to respond to advances in genetic science in the interpretation of human rights.¹²¹

4.3 The Universal Declaration on Bioethics and Human Rights

In 2005 UNESCO adopted the Universal Declaration on Bioethics and Human Rights,¹²² which has been described as “*an important step in the search for global minimum standards in biomedical research and clinical practice.*”¹²³ Against a clear human rights law framework, the primary aim was to provide basic provisions to ensure the appropriate use and application of biomedical research and technology.¹²⁴ This Declaration provides for a number of substantive principles in relation to bioethics, for example, respect for human dignity and human rights,¹²⁵ privacy and confidentiality,¹²⁶ equality, justice and

¹¹⁸ IDHGD, Article 14

¹¹⁹ IDHGD, Article 7

¹²⁰ IDHGD, Article 4

¹²¹ For further discussion see, Henriette Roscam Abbing, ‘International Declaration on Human Genetic Data’ (2004) 11 European Journal of Health Law 1 93

¹²² Universal Declaration on Bioethics and Human Rights adopted by UNESCO’s General Conference on 19 October 2005 (“UDBHR”)

¹²³ Roberto Andorno, ‘Global Bioethics at UNESCO: in defence of the Universal Declaration on Bioethics and Human Rights’ (2007) 33 Journal of Medical Ethics 150, 150

¹²⁴ Ibid at 153

¹²⁵ UDBHR, Article 3(1)

¹²⁶ UDBHR, Article 9

equity,¹²⁷ non- discrimination and non- stigmatisation.¹²⁸ The Declaration thereby reaffirms some of the key principles set out in the 1997 and 2003 Declarations. Although no clear regulatory framework is evident from the 2005 Declaration, it highlights the general legal and ethical considerations arising in these circumstances. It further highlights the evolution in the orientation of human rights and the recognition by the UN of the need to respond to advances in science and technology.

4.4 Other international documents

In 2004 the United Nations Economic and Social Council adopted a Resolution, 2004/9 on Genetic Privacy and Non- Discrimination.¹²⁹ This Resolution “[u]rges states to ensure that no one shall be subjected to discrimination based on genetic information” and to take the appropriate measures to attain this goal. This Resolution refers to the 1997 and 2003 Declarations, as well as core human rights documents such as the Universal Declaration of Human Rights. In terms of framing the main issues arising, the 2004 Resolution reaffirms the principles of genetic privacy and discrimination, and the potential for stigmatisation on the basis of one’s genes. In line with the other UNESCO Declarations, the 2004 Resolution emphasises the importance of human dignity and respecting fundamental human rights, while at the same time acknowledging the importance of advances in genetic science. This document contributes towards shaping the UN position in this area, and has a certain expressive value in elucidating the key ethical issues.

4.5 Evaluation of the UNESCO Declarations

In providing an international frame in this area, it has been noted that these instruments “... provide clear evidence of the importance of human rights as a jumping-off point in the evolving body of international law and practice on the regulation of new technologies.”¹³⁰ The Declarations represent an international commitment to ensuring that genetic science advances with appropriate safeguards and acknowledgement of the importance of human rights. The Declarations enhance our understanding of the area of bioethics and human rights, by highlighting the ethical contours of the debate and in providing a framework that is reflective of core human rights.

It has been observed that the instruments of UNESCO have been “a stimulus for national legislation in many countries throughout the world even though they are

¹²⁷ UDBHR, Article 10

¹²⁸ UDBHR, Article 11

¹²⁹ UNESCO Resolution 2004/9

¹³⁰ Therese Murphy, ‘Taking Revolutions Seriously: Rights, Risk and New Technologies’ (2009) 16 Maastricht Journal 21

not legally binding nor can they be ratified as such."¹³¹ In terms of highlighting a regulatory framework within which to view this area, the measures condemn discrimination on genetic grounds, emphasise that genetic information should be confidential and highlight the importance of maintaining human dignity. Although the guidelines are helpful in fostering an attitude against misuse of genetic information, the instruments are not legally binding. Nevertheless, despite the obvious drawbacks, the degree of consensus reached in this sensitive area is certainly admirable.

The symbolic significance of these Declarations is also recognised. As well as having a strong expressive value, the UNESCO Declarations constitute an initial, but significant endeavour towards regulation of genetic science and other technologies.¹³² In this regard, the rationale or function of a declaration is to generate harmony or agreement in a particular area, at an international level.¹³³ It is further observed that these declarations constitute a type of endeavour "*that is both psychological and political.*"¹³⁴ The values and principles espoused in these instruments have clear expressive value and can act as an impetus for discussion and action in a particular area. The underlying intention with these instruments was clearly to integrate the fundamental principles that arise in light of advancing science and to foster a certain degree of uniformity.

It is important to be aware of these international instruments and their objective in terms of setting a vital ethical standard and recognising the legal dilemmas arising, from a human rights perspective. It is clear from the above analysis that the majority of the international documents in this area reflect a clear human rights framework, since they generally prohibit differential treatment of individuals on the basis of genetic information, with a focus on the principles of human dignity¹³⁵ and equality.¹³⁶ More specifically, a common thread running through the Declarations is non- discrimination and to a certain extent, privacy. Although there are obvious shortcomings in terms of the non- binding and relatively broad nature of these instruments, nevertheless, they can potentially have a significant persuasive impact upon legislative endeavours.¹³⁷ Accordingly, we must be aware of the content of these instruments in considering the way forward in

¹³¹ Sirpa Soini, 'Genetic testing legislation in Western Europe – a fluctuating regulatory target' (2012) 3 Journal of Community Genetics 143, 145

¹³² Souheil El- Zein, 'Genetic Manipulation: How to Strike the Right Balance between Technology and Respect for Human Rights', in Antonio Cassese (ed) *Realizing Utopia- The Future of International Law* (Oxford University Press 2012) 485

¹³³ Noelle Lenoir, 'Universal Declaration on the Human Genome and Human Rights: The First Legal and Ethical Framework at the Global Level' (1998- 1999) 30 Columbia Human Rights Law Review 537, 549- 550

¹³⁴ Ibid at 551

¹³⁵ For further discussion on the notion of human dignity in this area, see Roberto Andorno, 'Global Bioethics at UNESCO: in defence of the Universal Declaration on Bioethics and Human Rights' (2007) 33 Journal of Medical Ethics 150, 153

¹³⁶ Yann Joly, Maria Braker and Michael Le Huynh, 'Genetic discrimination in private insurance: global perspectives' (2010) 29 New Genetics and Society 4 351, 357

¹³⁷ Ibid at 357

regulating the area of genetic information against the backdrop of a clear human rights framework.

5. Conclusion

As has been illustrated in this chapter, international human rights law is deeply rooted in principles of dignity, integrity and respect for human diversity. The first part of this chapter looked at the UN human rights treaties. It highlighted the relevance of the ICCPR and the ICESCR, in providing a clear human rights based approach towards recognising and guaranteeing a wide range of civil and political rights, as well as economic, social and cultural rights to all individuals including persons with disabilities. These rights reflect further fundamental human rights such as human dignity and self-determination, as well as facilitate an individual's fruitful participation in society. On examining UN treaty law, the second part of this chapter focused on the CRPD, as a key disability rights instrument in this area. Set against the backdrop of a clear social model approach to disability, the CRPD primarily utilises a non-discrimination framework to fulfill the rights of persons with disabilities. It is submitted that the CRPD has the potential to shape the regulation of genetic information at EU level. It certainly has the potential to inform the debate and provide a platform for discussion of the issues arising in this area, from a disability and rights based perspective.

The CRPD *“represents the more complex and layered human rights law appropriate to the challenges of the twenty-first century.”*¹³⁸ One such challenge of the twenty-first century is the reality of advancing genetic science and the question of regulating genetic information. These challenges give rise to ethical and legal dilemmas and a need to consider a balance between various competing rights. The question arises, to what extent can the CRPD provide a platform for discussion of this issue, and to what extent it can provoke and perhaps initiate legislative endeavours by the EU (particularly in light of the EU conclusion of the CRPD). Although entering new territory, and although there appears to be no legal obligations on the EU to take active steps to enact legislation, it is acknowledged that EU legislative endeavours in the area of disability non-discrimination and specifically genetic non-discrimination may well be the preferable way forward.

This chapter also provided an overview of the UN soft law instruments in this area. These Declarations emphasise a prohibition on genetic based discrimination, as well as highlight the necessity of respecting privacy and confidentiality. Although, these soft-law instruments have been influential in providing a clear response to the area of bioethics and genetics in particular, and

¹³⁸ Oddný Mjöll Arnardóttir, 'A Future of Multidimensional Disadvantage Equality?' in Oddný Mjöll Arnardóttir and Gerard Quinn (eds) *United Nations Convention on the Rights of Persons with Disabilities* (Martinus Nijhoff Publishers 2009) 66

admirably placing fundamental human rights at the forefront, their practical application is limited. These declarations nevertheless provide a framework within which to consider the ethical and legal dilemmas arising with the advent of advancing genetic science and technology. They also reflect the clear evolution in human rights and their interpretation.

The following chapter will analyse the comparative legislative benchmarks in the area, by looking at the position in the US and Australia.

Chapter 7: Evolving Comparative Law Benchmarks

1. Introduction

This chapter will explore the response to the issue of advancing genetic technology in the US and Australia. The choice was made to look at these two countries, as these are two of the most developed jurisdictions, from a legislative and policy perspective. There has also been a significant amount of academic literature generated from these jurisdictions that has further shaped the interpretation of the issues in this area. In addition, it is acknowledged that the majority of legislative and policy endeavours in these countries have taken place at federal level, which is useful in terms of considering EU level regulation in this area.¹ Accordingly, it is interesting to examine how these jurisdictions have grappled with the ethical and legal issues arising and look to how they have conceptualised the framework of addressing use and misuse of genetic information. Although similar issues arise, this chapter also acknowledges some differences in the social and historical backgrounds of these jurisdictions.

This chapter will firstly look at the position in the US, which adopted specific federal level legislation on the issue of genetic non- discrimination in employment and health insurance. In terms of a regulatory framework, it has chosen a clear non- discrimination approach. It has also favoured the merits of a stand- alone genetic specific response. This section will examine the history behind this legislative endeavour and will evaluate the effectiveness of the law.

This chapter will also outline the response of Australia in this area. In contrast to the US, Australia's response to this area can be characterised primarily by a major national level empirical study of genetic discrimination, as well as an investigation by the Law Reform Commission, which made recommendations in this area. Australia decided to amend existing legislation, as opposed to enacting new legislation to address misuse of genetic information. It addressed the issue from a non- discrimination, as well as a privacy approach. These comparative perspectives and benchmarks highlight the legislative and policy endeavours taking place, and contribute towards shaping the debate in the EU.

¹ However, it is acknowledged that the EU is not a federal state. It has been described as a "staatenverbund": an association of sovereign states. See Elisabetta Layza, 'Core of State Sovereign and Boundaries of European Union's Identity in the *Lissabon – Urteil*' (2010) 11 German Law Journal 399

2. United States

This chapter considers the US position, on account of the US being a leader in introducing specific federal legislation in this area, and also in recognition of the influence of US law and policy in the EU.² When considering the regulation of genetic information at EU level, it is useful to examine transatlantic perspectives on the matter, taking into account the importance of the relationship between the US and the EU, as well as the objective of building upon this relationship when engaging in law and policy reform.³ Both jurisdictions are dealing with similar ethical and legal dilemmas stemming from advancing genetic technology, albeit against the backdrop of different historic and political settings.

This section will highlight the Genetic Information Nondiscrimination Act 2008 (GINA), a federal law that targets genetic discrimination by employers and health insurers.⁴ A few observations about GINA can be made from the outset. Drafters of the legislation recognised that genetic information is unique and merits special protections.⁵ In terms of a regulatory framework, GINA is a civil rights law and an anti-discrimination framework was favoured as an approach. The legislation is novel in the US in that it is a departure from every anti-discrimination statute preceding it. It is the first preemptive anti-discrimination law in US history, meaning that it attempts to eliminate a new type of discrimination before it becomes widespread.⁶ This can be contrasted with previous civil rights laws in the US, which generally look for a documented history of discrimination as the basis to prohibit similar conduct in the future.⁷

This section will examine the reasons behind GINA and the opposition faced. It will examine the balance struck in GINA, by analysing the key provisions and exceptions contained therein. Finally, the section will evaluate the effectiveness

² See generally Gerard Quinn and Eilionoir Flynn, 'Transatlantic Borrowings: The Past and Future of EU Non-Discrimination Law and Policy on the Ground of Disability' (2012) 60 *American Journal of Comparative Law* 23. See also Gerard Quinn and Aisling de Paor, 'The New Transatlantic Agenda and the United Nations Convention on the Rights of Persons with Disabilities: Taking the US/EU Dialogue to a New Level' (2010: US International Council on Disability, Washington D.C.), available at <http://www.usicd.org/doc/10%2001%2015%20The%20New%20Transatlantic%20Agenda%20and%20the%20UNCRPD%20AP%20RevisedVersion.pdf> (accessed 11 April 2012). See Michael A. Stein et al, 'The Law and Politics of US Participation in the UN Convention on the Rights of Persons with Disabilities', in Gerard Quinn and Lisa Waddington (eds) 2 *European Yearbook of Disability Law* (2010 Intersentia). See also Aart Hendriks, 'Genetics, human rights and employment. American and European Perspectives' (1997) 16 *Medicine and Law* 557

³ *Ibid*

⁴ Genetic Information Nondiscrimination Act of 2008, Pub. L. No. 110-233, 122 Stat. 881 (2008) ("GINA")

⁵ See generally Thomas H. Murray 'Genetic Exceptionalism and "Future Diaries": Is Genetic Information Different from Other Medical Information?' in Mark A. Rothstein (ed) *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic* (Yale University Press 1997)

⁶ Jessica L. Roberts, 'Preempting Discrimination: Lessons from the Genetic Information Nondiscrimination Act' (2010) 63 *Vanderbilt Law Review* 2 439, 441

⁷ Americans with Disabilities Act of 1990, Pub. L. No. 101-336 (1991) ("ADA")

of GINA as a regulatory regime. Before exploring GINA, it is necessary to provide a historical context to illustrate the backdrop against which GINA was advocated and introduced.

2.1 Differences between the insurance industry and role of insurance in US and Europe

It is important to emphasise that there are differences between the insurance industry and role of insurance in the US and the EU. There is currently no universal health care provided by the government in the US, although this is currently under reform (noted below). Therefore health insurance is generally provided for by private health insurers. In addition, the majority of individuals in the US obtain insurance cover through their employers. Therefore access to health care is a serious problem, particularly for the unemployed. Insurance is therefore closely linked to employment in the US.

Employers in the US have an even greater incentive than employers in other countries to exclude employees who are or who may become ill in the future.⁸ In addition, health insurance obtained through employers often extends to cover dependents, therefore employers have further reason to discriminate against employees who have dependents who are ill or who are likely to become ill.⁹ The US position is in comparison to most EU countries, which have some form of universal access to health care. In spite of the fundamental differences between these two jurisdictions, both are dealing with similar challenges in this area.

2.2 Historical background of eugenics in the US

In chapter 3, the historical background of eugenics and abuse of science in the US was identified and discussed. Historical incidents of eugenics and discrimination appear to have scarred the minds of individuals and contributed towards the widespread fear of genetic discrimination that is evident in the US.¹⁰ Indeed such fear was one of the reasons cited in advocating the need for genetic information non-discrimination legislation.¹¹ The historical and social background of genetics-based eugenics set the context for the modern day fear of genetic discrimination that is still entrenched in American society.

⁸ Mark A. Rothstein, Bartha Maria Knoppers, 'Legal Aspects of Genetics, Work and Insurance in North America and Europe' (1996) 3 *European Journal of Health Law* 134, 144

⁹ *Ibid*

¹⁰ Martin S. Pernick, *The Black Stork: Eugenics and the Death of "Defective" Babies in American Medicine and Motion Pictures Since 1905* (Oxford University Press 1996)

¹¹ GINA, s 2(5)

It has been documented that the poor, the ostracised, and those perceived as disabled have long been the targets of eugenic policies.¹² These incidences of stigmatisation, eugenics and genetic determinism highlight historical patterns of prejudice and discrimination in the US, that provide a historical and societal context to the introduction of GINA. This historical background certainly informed the debate in the US and served to highlight the core concerns in this area, from a societal perspective. In addition, such historical patterns of prejudice and discrimination in the US raise the question whether the benefits of genetic technology can be realised in a way that avoids introducing a new era of social oppression. Such considerations further point to the need for appropriate protections in this area, in the absence of which there is a real potential for further development of eugenic practices.

2.3 Legislative history

Having highlighted the historical and cultural backdrop within which to understand and frame the discussion in the US, the following section will give an overview of the legislative history and the reasons behind GINA. GINA is the result of more than two-dozen proposed bills and underwent a 13- year journey through Congress, encountering much opposition. When the first federal legislation to prevent the misuse of genetic information was introduced in 1995, many stakeholders viewed the law as being forward looking, while others called it “*premature*.”¹³ At this time, scientists were still making genetic discoveries and the Human Genome Project had not yet been completed. In addition, there were not many genetic tests available, and the tests were generally unavailable to many. However, science and technology were moving at a rapid pace and the potential misuse of this new technology was also becoming apparent.

2.3.1 Evidence of genetic discrimination and fear of genetic discrimination

Prior to the introduction of GINA, there was only limited evidence indicating the extent of the actual discrimination. Accordingly, there was a limited amount of empirical data (to verify the reality of misuse of genetic information or fear of discrimination) in the US. However, some cases of actual and anecdotal evidence of genetic discrimination had been compiled. By August 1997, the Council for Responsible Genetics had documented over 200 cases of genetic discrimination.¹⁴ These cases documented a range of discrimination on the basis of genetic information by insurance companies, employers and others, against asymptomatic individuals with genetic predispositions to certain conditions.

¹² For further discussion, see Kenneth Ludmerer *Genetics and American Society: a historical appraisal* (Johns Hopkins Press, 1972), Daniel Kevles *In the Name of Eugenics: Genetics and the Uses of Human Heredity* (University of California 1985)

¹³ Kathy L. Hudson et al, ‘Keeping Pace with the Times – the Genetic Information Nondiscrimination Act of 2008’ (2008) 358 *New England Journal of Medicine* 2661, 2662

¹⁴ See: <http://www.councilforresponsiblegenetics.org/> (12 May 2013)

Therefore, even at this stage, there was an indication that reform of the law in this area was becoming an increasing necessity.

During Congressional testimony, reference was made to the reality of genetic discrimination and the various surveys and studies that had been carried out. In 1998, the National Center for Genome Resources (NCGR) carried out a national survey to assess the public's attitude toward genetic concerns.¹⁵ The NCGR found that 85 percent of those surveyed believe employers should be prohibited from obtaining genetic information about an individual's genetic conditions and predispositions to disease. The study also highlighted that 36 percent of those surveyed were not likely to take genetic tests and 27 percent would definitely not take such tests if health insurers or employers had a right to access the test results, thereby indicating a fear of discrimination or misuse.¹⁶

Congressional testimony also noted an interagency report that was jointly issued by the Equal Employment Opportunity Commission (EEOC) and the Departments of Labor, Health and Human Services and Justice in 1998.¹⁷ The report stated that, “[f]ederal legislation is needed to ensure that knowledge gained from genetic research is fully utilized to improve the health of Americans and not to discriminate against workers.”¹⁸ The report therefore recognised the gaps in protection and supported the need for a federal level response.

The history of the eugenics movement in the US, together with the reported fear of genetic discrimination and anecdotal evidence highlighted, provided motivation for introducing a clear framework at federal level. The following section will look at some further reasons for GINA and some of the opposition faced.

2.4 Reasons behind GINA and opposition faced

On examination of the Congressional testimony, it is evident that GINA was introduced in response to scientific developments that proponents worried may lead to a unique type of discrimination, in the absence of legal protections.¹⁹ During Congressional testimony, Francis Collins emphasised the endeavours of scientists and the success of the Human Genome Project.²⁰ He focused on the

¹⁵ The National Center for Genome Resources survey, ‘Attitudes Toward Genetic Testing’ (4 March 1998)

¹⁶ Ibid

¹⁷ Interagency report: ‘Genetic Information and the Workplace,’ (Department of Labor, Department of Health and Human Services, Equal Employment Opportunity Commission, Department of Justice,’ 20 January 1998) [http://dol.gov/dol/_sec/public/media/reports/genetics.htm]

¹⁸ Ibid

¹⁹ Jessica L. Roberts, ‘Preempting Discrimination: Lessons from the Genetic Information NonDiscrimination Act’ (2010) 63 Vanderbilt Law Review 2 439, 442

²⁰ Statement of Francis S. Collins, (Director, National Human Genome Research Institute), before the Appropriations Subcommittee on Labor, Health and Human Services and Education (11 July 2001) See also statement of Francis S. Collins, before the Health, Education, Labor and Pensions Committee, United States Senate (25 July 2001)

great promise that genetic science holds for all individuals in terms of treatments and disease prevention and generally improving public health.²¹

Linked to this, it was also argued that specific genetic information legislation was needed to alleviate the fear of genetic discrimination that was preventing some individuals from undergoing genetic testing that may improve their health.²² The undesirable impact of this fear on individual and public health is noted.²³ Proponents of protecting genetic information argued that the fear of genetic testing was negatively impacting genetic research. In order for genetic science to advance, clinical studies and trials are required. However, fear of discrimination deters individuals from undergoing genetic testing and participating in studies. This consequently inhibits research and stifles genetic innovation.²⁴ A further reason for enacting GINA was to address the concern that a new type of eugenics will develop.²⁵ There was a fear that genetic discrimination could be used as a “*proxy*” for discrimination on the grounds of race,²⁶ with particular reference to the controversial history of eugenics and the adverse treatment of African- Americans in the 1970s, as previously illustrated.

One of the main arguments made by opponents was the lack of widespread genetic discrimination. Opponents maintained that there was little evidence that employers and insurers were engaging in discrimination on the basis of genetic information. GINA's opponents noted the absence of genetic discrimination as an indication that the law was not necessary.²⁷ Indeed, at the time, the legislative initiatives were deemed “*a remedy in search of a problem*.”²⁸ Conversely, GINA's advocates also cited the lack of widespread discrimination, and viewed this as an opportunity to preemptively legislate and adopt a forward- thinking approach,

²¹ Ibid

²² Jessica L. Roberts, ‘The Genetic Information Nondiscrimination Act as an Antidiscrimination Law’ (2011) 86 Notre Dame Law Review 2 597, 605

²³ Francis Collins, ‘The Threat of Genetic Discrimination to the Promise of Personalized Medicine’, Testimony before the Subcommittee on Health Committee on Energy and Commerce, United States House of Representatives (9 March 2007) Available at: <http://www.genome.gov/Pages/Newsroom/SpeechesAndTestimony/CollinsGINATestimony030807.pdf> (accessed 18 April 2012)

²⁴ Press Release: ‘Landmark Snowe Bill to Ban Genetic Discrimination passes Senate’ (24 April 2008). Available at http://snowe.senate.gov/public/index.cfm/pressreleases?ContentRecord_id=81ffaa24-802a-23ad-497c-c4bc27b9941b&ContentType_id=ae7a6475-a01f-4da5-aa94-0a98973de620&Group_id=2643ccf9-0d03-4d09-9082-3807031cb84a&MonthDisplay=4&YearDisplay=2008 (accessed 17 April 2012)

²⁵ Shannyn C. Riba, ‘The Use of Genetic Information in Health Insurance: Who will be Helped, Who will be Harmed and Possible Long-Term Effects’ (2007) 6 Review of Law and Social Justice 2 469, 487

²⁶ Ibid at 487

²⁷ Jessica L. Roberts, ‘Preempting Discrimination: Lessons from the Genetic Information Nondiscrimination Act’, (2010) 63 Vanderbilt Law Review, 2 439, 441

²⁸ Statement of Burton J. Fishman, ‘Protecting Workers from Genetic Discrimination:’ Hearing Before Subcommittee on Health, Employment, Labor and Pensions of the Committee on Energy and Commerce, 110th Congress 110-1 (30 January 2007)

before it has the opportunity to become problematic.²⁹ Another reason behind GINA was the patchwork of protections that was evident at the state and federal level.

2.5 Legislative position prior to GINA – a limited patchwork

Prior to GINA, there was effectively a patchwork of varying legislation, with states introducing their own protections.³⁰ There was also a patchwork of federal protections that may be interpreted to encompass genetic information discrimination, but none of which provided comprehensive legislative protection. This section will give a brief overview of the relevant federal level legislation, prior to GINA.

Firstly, Title VII of the Civil Rights Act 1964 contains protections on a number of grounds (including race, religion and sex) in the employment context.³¹ There is no reference to discrimination on the basis of genetic information, however, in light of the racial and ethnic links with certain genetic conditions, an argument could be made that genetic discrimination may constitute unlawful race discrimination, or sex discrimination. A relevant case under this legislation is *Norman- Bloodsaw v Lawrence Berkeley Laboratory*.³² In this case, the main claim came under Title VII, whereby allegedly an employer required its black and female employees to undergo testing for sickle cell trait, syphilis, and pregnancy. The Court held that the employees' claims under Title VII were valid, as the discriminatory acts of the employer specifically targeted race and sex. Arguably if there had been genetic information non- discrimination legislation at federal level legislation at this time, the employer would have been prohibited from gathering this genetic information.³³

The Americans with Disabilities Act (ADA) 1990³⁴ and the Americans with Disabilities Amendment Act 2008³⁵ provide protections against disability discrimination. There was uncertainty as regards the scope and application of some of the existing protections, for example as regards the application of the ADA to cover genetic information. The application of the ADA to the issue of

²⁹ Jessica L. Roberts, 'Preempting Discrimination: Lessons from the Genetic Information Nondiscrimination Act' (2010) 63 Vanderbilt Law Review 2 439, 441

³⁰ For example, in 1991, Wisconsin was the first state to introduce legislation prohibiting health insurers from requesting genetic information or using such information. Since then, most states have introduced legislation regulating genetic information. Such a patchwork is currently developing in the EU (discussed in chapter 10)

³¹ Title VII of the Civil Rights Act of 1964 Pub. L. No. 88 – 352 (1964)

³² *Norman- Bloodsaw v Lawrence Berkeley Laboratory*, 135 F.3d 1260, 1265 (9th Cir. 1998) at 1271-72.

³³ For further discussion see Daniel Schlein, 'New Frontiers for Genetic Privacy Law: The Genetic Information NonDiscrimination Act of 2008' (2008- 2009) 19 Civil Rights Law Journal 2 318, 329

³⁴ ADA

³⁵ Americans with Disabilities Amendment Act of 2008 Pub. L. No. 110-325 (2008)

genetic information or genetic predisposition to disability was contentious and gave rise to diverging opinions, policies and judicial interpretation.³⁶

In 1995, the EEOC issued an interpretation stating that the ADA prohibits discrimination against employees based on their genetic make up.³⁷ Even though the EEOC came out with a firm stance in favour of the ADA protecting individuals with genetic markers against discrimination in employment, this interpretation does not have the same value as a federal law. This point was referred to in Congressional testimony.³⁸ In addition, although the EEOC interpretation was welcomed, it was further limited in application following a series of decisions of the Supreme Court on the definition of disability.³⁹ Indeed during Congressional testimony for GINA, the requirement for specific legislation to clarify the prohibition against discrimination based on genetic predisposition was further reinforced given the US Supreme Court's decisions interpreting the ADA.

Outside the employment context, the Health Insurance Portability and Accountability Act, 1996 (HIPAA)⁴⁰ governs whether and in what manner group health insurers may obtain or consider genetic information in health insurance coverage. HIPAA prohibits group health insurers from using genetic information to vary premiums, limit an individual's eligibility for insurance, or deny insurance coverage. Arguably, HIPAA is an important first step in tackling genetic discrimination in health insurance. However, despite the protections afforded, HIPAA fails to prohibit several types of genetic discrimination. It also fails to cover individual health insurance plans.⁴¹

Federal employees in the US have had far more extensive protections than private sector employees pursuant to Executive Order 13145,⁴² which governs the use of genetic information by executive departments and agencies. The Executive Order explicitly prohibits discrimination on the basis of protected

³⁶ For further discussion see Daniel Schlein, 'New Frontiers for Genetic Privacy Law: The Genetic Information NonDiscrimination Act of 2008' (2008- 2009) 19 Civil Rights Law Journal 2 318, 319-327. See also Deborah Kaplan, 'The Definition of Disability: Perspective of the Disability Community' (1999- 2000) 31 Health Law and Policy 352

³⁷ EEOC Compliance Manual, Volume 2 EEOC Order 915.002. Definition of the concept "disability" at 902- 45

³⁸ Statement of Paul Steven Miller on Genetic Information in the Workplace, US Equal Employment Equality Commission before the Committee on Health, Education, Labor and Pensions, US Senate (20 July 2000)

³⁹ On 22 June 1999 the Supreme Court handed down a series of decisions which restricted the interpretation of the coverage of the ADA. These key cases were: *Sutton v United Air Lines Inc.*, 119 S. Ct 2139 (1999),

Murphy v United Parcel Service Inc. 119 S. Ct 2133 (1999) and *Albertsons Inc. v Kirkingburg* 119 S. Ct. 2162 (1999)

⁴⁰ Health Insurance Portability and Accountability Act of 1996 Pub. L. No. 104- 191 (1996)

⁴¹ For further discussion see Daniel Schlein, 'New Frontiers for Genetic Privacy Law: The Genetic Information NonDiscrimination Act of 2008' (2008- 2009) 19 Civil Rights Law Journal 2, 318, 336-337

⁴² Executive Order No. 13145, 65 Fed. Reg. 6,877 (2000)

genetic information in all aspects of employment in executive departments and agencies and limits federal employers' access to, and use of genetic information. However, although welcomed protections, the scope of this Executive Order is limited to federal employees.⁴³

Prior to GINA there were a number of cases indicating the potential of genetic discrimination, particularly in the employment context. For example, a seminal case is *EEOC v Burlington Northern Santa Fe Railroad*.⁴⁴ In this case, the EEOC alleged unlawful genetic discrimination and settled its first court action challenging the use of genetic testing in the workplace under the ADA against the Burlington Northern Santa Fe Railroad (BNSF). The BNSF tested employees to identify a genetic marker for carpal tunnel syndrome to address a high incidence of repetitive stress injuries among its employees. At least one employee had been threatened with discipline and possible termination for refusing to take the test. The EEOC found that the alleged genetic discrimination had violated the disability discrimination laws. As a settlement was agreed for the sum of \$2.2 million out of court on 18 April 2001, no court ruled on the applicability of the disability discrimination theory.⁴⁵ However, the case highlighted the reality of the issue of genetic discrimination and the implications in the employment context.

2.6 The arrival of GINA

Accordingly, in light of the varying levels of protection, an element of the intention behind GINA was therefore to “*eliminate existing gaps in coverage and ambiguities in the disparate statutes, administrative interpretations, and regulations that affect health insurance coverage and employment rights*.”⁴⁶ The fragmented federal and state protection and the gaps in protection compounded the need for comprehensive federal level legislation. GINA was introduced on 21 May 2008⁴⁷ and was enacted by Congress on the basis of the Commerce Clause. The Commerce Clause⁴⁸ grants Congress the power to regulate interstate commerce. It was the case that the existing patchwork of laws in the US inhibited the operation of interstate commerce. Employment and insurance will often significantly influence interstate commerce and the economy, thereby justifying federal level action, rather than state action.

⁴³ Deborah Josefson, 'Clinton outlaws genetic discrimination in federal jobs', (2000) 320 *British Medical Journal* 723 468

⁴⁴ Patricia Roche, 'The Genetic Revolution at Work: Legislative Efforts to Protect Employees' (2002) 28 *American Journal of Law and Medicine* 271, 272

⁴⁵ Press Release, EEOC, 'EEOC Settles ADA Suit Against BSNF for Genetic Bias (18 April 2001) available at: <http://www.eeoc.gov/press/4-18-01.html> (accessed 26 July 2013)

⁴⁶ Daniel Schlein, 'New Frontiers for Genetic Privacy Law: The Genetic Information NonDiscrimination Act of 2008' (2008- 2009) 19 *Civil Rights Law Journal* 2 311, 313

⁴⁷ GINA. For further discussion, see Jessica Roberts, 'The Genetic Information Non Discrimination Act as an Antidiscrimination Law' (2011) 86 *Notre Dame Law Review* 2 597

⁴⁸ Constitution of the United States, Article 1, section 3, clause 3

Senator Edward Kennedy referred to GINA as “*the first civil rights bill of the new century*”, and it was hailed as a milestone in the protection of the most personal information.⁴⁹ The law was enacted to help eliminate worries about discrimination that might dissuade individuals from taking advantage of genetic technologies that could improve their health.⁵⁰ The law also enables people to take part in research studies without fear that their genetic information might be used against them.⁵¹ The objective to enhance public confidence was clear in the legislation. The following section will give an overview of the main provisions and exceptions in GINA.

2.6.1 GINA – main provisions

GINA is divided into three different titles. Firstly, there is an extensive Preamble, which is illustrative of the context within which the legislation was introduced, and provides an insight into the US’s history of genetic discrimination as well as the social and political background. It refers to the technological advances made, the connection between genetic discrimination and racial discrimination, and the current need for protections for genetic information.⁵²

Specifically, Title I targets genetic information discrimination in health insurance. Title I prevents health insurers from using genetic information to determine coverage, eligibility or premiums, and prevents insurers from requesting or requiring genetic testing or genetic information for underwriting purposes.⁵³ Title II applies to genetic discrimination in employment.⁵⁴ Title II makes it unlawful for employers to make hiring and termination decisions on the basis of employees’ genetic information.⁵⁵ Employers are prevented from using genetic information to discriminate against the employee regarding compensation, benefits, or other terms and conditions of employment.⁵⁶ An employer is also prohibited from requesting, requiring, or purchasing genetic information of an employee or from family members of the employee.⁵⁷ Title I and Title II therefore contain relatively

⁴⁹ Press Release: Kennedy, Enzi, Snowe Celebrate Passage of Genetic Information NonDiscrimination Act (24 April 2008), available at:

<http://www.help.senate.gov/newsroom/press/release/?id=313bfde8-f967-46b4-aa9d-11bc73728813> (accessed 18 April 2012)

⁵⁰ Francis Collins, Testimony before the US House of Representatives Subcommittee on Health: Committee on Energy and Commerce on the Threat of Genetic Discrimination to the Promise of Personalized Medicine (March 8, 2007), available at:

<http://www.hhs.gov/asl/testify/2007/03/t20070308l.html> (accessed 18 April 2012).

⁵¹ For further discussion on the fear of genetic information discrimination, see Karen H. Rothenberg and Sharon F. Terry, ‘Before It’s Too Late – Addressing the Fear of Genetic Information’ (2002) 297 Science 196, 196. See also Jessica L. Roberts, ‘The Genetic Information Nondiscrimination Act as an Antidiscrimination Law’ (2011) 86 Notre Dame Law Review 2 597, 603

⁵² GINA, Preamble

⁵³ GINA, s 101- 106

⁵⁴ GINA, s 202

⁵⁵ GINA, s 202 (a)(1)

⁵⁶ GINA, s 202 (a)(1)

⁵⁷ GINA, s 202 (b)

strong protections against the misuse of genetic information in health insurance and employment.

GINA also provides for a Genetic Nondiscrimination Study Commission to be established by May 2015,⁵⁸ the objective of which is to review scientific advancements in genetics and advise Congress as to the potential need to amend GINA. It will provide a mechanism through which to monitor and keep up to date with advances in genetic science and additional legal and ethical issues that may arise.⁵⁹

It is noted that the legislation strikes a balance and also includes a number of exceptions which will be discussed in the following section.

2.6.2 GINA's Exceptions

GINA provides for a number of exceptions, representing the compromise made in an effort to ensure the passage of the legislation.⁶⁰ Title I contains a limited exception for a group insurer or issuer that wishes to obtain genetic data for research purposes.⁶¹ Health insurers are also not penalised for incidentally acquiring genetic information. Similarly, Title II does not apply to incidental acquisition of genetic information, for example, if an employer inadvertently acquires genetic information through casual conversation with an employee.⁶²

Another exception relates to the acquisition of information from purchasing commercially or publicly available documents that contain an employee's family medical history, such as an obituary in a newspaper.⁶³ Employers can therefore legitimately glean sensitive information about an employee's family history from family obituaries. These exceptions draw attention to the challenges that arise in limiting the flow of genetic information and represent a practical compromise in the regulation of genetic information. Kim makes several arguments as to why preventing employers from acquiring any genetic information about their employees will be difficult. In addition, further public policy concerns may dictate that employers have access to genetic information – for example, genetic monitoring to identify the workplace hazards.⁶⁴

⁵⁸ GINA, s 208(b)

⁵⁹ Joanne Barken, 'Judging GINA – Does the Genetic Information Nondiscrimination Act of 2008 offer Adequate Protection?' (2009-2010) 75 Brooklyn Law Review 546, 547

⁶⁰ Susannah Baruch and Kathy Hudson, 'Civilian and Military Genetics: Nondiscrimination Policy in a Post- GINA World' (2008) 83 American Journal of Human Genetics 435, 435

⁶¹ GINA section 101(b), 102(a), 103(b), 104(b)

⁶² GINA s 202 (b) (1)

⁶³ GINA s 202 (b) (4)

⁶⁴ Pauline Kim, 'Regulating the use of Genetic Information: Perspectives from the US Experience' (2010) 31 Comparative Labor Law and Policy Journal 4 693, 701- 702

A further exception is intended to promote the operation of employer-sponsored health or genetic services, including wellness programs and permits the relevant health professional to discuss genetics information with the employee.⁶⁵ Title II provides that the acquisition of genetic information by an employer is not unlawful if an employer offers a wellness program that includes genetic services, on the condition that the employee voluntarily participates and any individually identifiable information is not disclosed to the employer. This exception is open to criticism however, and it may operate to disadvantage employees and potentially result in adverse treatment of an employee. This exception represents a compromise between protecting employees' genetic information, promoting the wellbeing of a workforce and ultimately attempting to reform the overall health of the general population.

In striking a balance between the rights of employers and employees and in addressing the various concerns relating to health and safety, GINA permits an employer to undertake genetic monitoring to determine the biological effects of exposure to toxic substances in the workplace, but only under strict conditions (to ensure that an employee's rights are considered and upheld).⁶⁶ This exception is important in acknowledging the rights of employers and employees, and recognising an employer's obligations in hazardous workplaces.

It is important to note that when an employer or health insurer obtains genetic information under any of the exceptions, the information cannot be used to discriminate against the employee or insured and the confidentiality of the information must be maintained.⁶⁷ However, if genetic information gets into the hands of an employer or health insurer, it might be impossible to stop it from influencing decisions, to a certain extent anyway.⁶⁸ Kim puts forward the view that the exceptions to the prohibition on obtaining genetic information will make it more challenging to achieve the goal of *"fully protecting the public from discrimination and allaying their concerns about the potential for discrimination."*⁶⁹ The exceptions may therefore operate to dilute the effectiveness of the protections.

2.7 Cases under GINA

There have been very few cases taken under GINA. This section will highlight two recent cases that the EEOC has filed in respect of genetic information violations under GINA. The first suit filed by the EEOC was against a fabric

⁶⁵ GINA, s 202 (b) (2) (A)

⁶⁶ GINA, s 202 (b) (5). See also GINA, s 202 (b) (A), (B), (C)

⁶⁷ GINA, s 202 (c)

⁶⁸ Pauline Kim, 'Regulating the use of Genetic Information: Perspectives from the US Experience' (2010) 31 Comparative Labor Law and Policy Journal 693, 702

⁶⁹ Ibid.

distributor, Fabricut Inc.⁷⁰ The EEOC charged that Fabricut Inc. violated both the ADA and GINA. It was alleged to have violated the ADA by refusing to hire the individual by regarding her as having Carpal Tunnel Syndrome. It was alleged to have violated GINA in terms of requesting family medical history at the post- offer medical assessment. The case was filed and settled on the same day on 7 May, 2013, in U.S. District Court for the Northern District of Oklahoma.⁷¹ Fabricut agreed to a \$50,000 in settlement. In addition, it agreed to take certain actions to prevent such prohibited action in the future, to include the provision of anti-discrimination training to employees, the distribution of anti- discrimination policies to employees as well as notifying employees of the anti- discrimination position.⁷²

The second case filed by the EEOC was a class action against Founders Pavilion Inc. In this case, the EEOC alleged violation of the ADA, Title VII of the Civil Rights Act, and GINA.⁷³ In relation to the violation of GINA, Founders Pavilion Inc carried out post- offer, pre- employment medicals of applicants, including requesting family medical history. The EEOC first tried to come to a settlement and subsequently filed the case in the US District Court for the Western District of New York in Rochester.⁷⁴ The fact that there have been very few cases taken may be an illustration of the success of the legislation in deterring and preventing misuse of genetic information. It might also be the case that there is a lack of awareness of the potential of genetic science and the issue of genetic discrimination amongst the public and amongst interested parties such as employers and insurance companies. Perhaps there will be more cases taken under GINA as individuals become more aware of their rights and the legislative protections in place. Indeed, these recent cases taken may act as an impetus for further cases in this area.

2.8 Evaluating GINA

Although GINA is a welcomed contribution to the civil rights movement and represents a forward- thinking approach towards regulating genetic information, it is not a perfect piece of legislation and certain shortcomings can be observed.

⁷⁰ EEOC Press Release, 'Fabricut to Pay \$50,000 to Settle EEOC Disability and Genetic Discrimination Lawsuit': <http://www.eeoc.gov/eeoc/newsroom/release/5-7-13b.cfm> (7 May 2013) (accessed 23 May 2013)

⁷¹ *Equal Employment Equality Commission v Fabricut Inc.* Case No.: 13-cv-248-CVE-PJC (filed 7/05/2013)

⁷² *Equal Employment Equality Commission v Founders Pavilion Inc.d/b/a Founders Pavilion* Case No. 6:13-cv-06250-CJS (filed 16/05/2013)

EEOC Press Release, 'Fabricut to Pay \$50,000 to Settle EEOC Disability and Genetic Discrimination Lawsuit': <http://www.eeoc.gov/eeoc/newsroom/release/5-7-13b.cfm> (7 May 2013) (accessed 23 May 2013)

⁷³ EEOC Press Release, 'EEOC Files Class Genetic Discrimination Suit Against Corning Rehab Center': <http://www.eeoc.gov/eeoc/newsroom/release/5-16-13a.cfm> (16 May 2013) (accessed 23 May 2013)

⁷⁴ Ibid.

First, the prohibition on genetic discrimination is arguably not comprehensive. It applies just to discrimination in health insurance and employment. The legislation does not apply to life insurance,⁷⁵ or other potential uses of genetic information (for example, mortgage protection insurance). In addition, GINA does not apply to members of the military, to veterans obtaining health care through the Department of Veterans Affairs, or to the Indian Health Service. Therefore, the scope of the protection is limited,⁷⁶ and it is ironic that GINA specifically states that federal legislation “*is necessary to fully protect the public from discrimination and allay their concerns about the potential for discrimination...*”⁷⁷ It is questionable whether the legislation “*fully*” protects individuals in light of the limited scope. In this regard, Rothstein offers the insight, “*to allay public concerns about genetic discrimination, it’s necessary to prohibit the adverse treatment of individuals in numerous settings.*”⁷⁸

These gaps in protection may impact upon an individual’s willingness to undergo genetic testing. As a result of the fear of third parties misusing genetic information in a range of uncovered settings, individuals may decide not to avail of genetic technologies. This also has a corresponding negative impact upon physicians trying to treat patients, as well as the wider scientific community.⁷⁹

It is therefore unclear whether GINA’s provisions are sufficient to reassure the public that their genetic information will be protected, as was the intention of the legislation. GINA therefore only provides partial protection against genetic discrimination. However, to include all the scenarios in which misuse of genetic information can occur would arguably be impractical and difficult to reach consensus on.

In addition, shortcomings can be observed with the employment provisions of GINA, which have been described as “*ineffective.*”⁸⁰ The main dilemma arising is the practical challenge of differentiating between genetic and non- genetic medical records. By virtue of the employment provisions in GINA, employers are prohibited from requiring or requesting an individual to undergo a genetic test. Employers are further prohibited from requiring or requesting that an employee disclose the results of a genetic test as a condition of employment.⁸¹ However, these provisions are, to an extent, ineffective, in light of the provisions of the

⁷⁵ Brianna E. Kostecka, ‘GINA will protect you, just not from death: the Genetic Information Nondiscrimination Act and its Failure to include Life Insurance within its Protections’ (2009- 2010) 34 Seton Hall Legislative Journal 94

⁷⁶ Amy L. McGuire and Mary A. Majumder, ‘Two Cheers for GINA?’ (2009) 1 Genome Medicine 6

⁷⁷ GINA, s 2 (5)

⁷⁸ Mark A. Rothstein, ‘GINA’s Beauty is Only Skin Deep’ (2009) 22 GeneWatch 2 9, 9

⁷⁹ Timothy J. Aspinwall, ‘Imperfect Remedies: Legislative Efforts to Prevent Genetic Discrimination’ (2010) 19 Annals of Health Law – 25th Anniversary Special Edition 121, 123

⁸⁰ Mark A. Rothstein ‘GINA’s Beauty is Only Skin Deep’ (2009) 22 GeneWatch 2 9, 10

⁸¹ Ibid

ADA,⁸² which make it lawful, following a conditional offer of employment, for an employer to require individuals to undergo a pre-placement medical examination and to authorise the release of all of their medical records/ information to the employer. At the moment, there is no practical means to segregate genetic information from non- genetic information.⁸³

Therefore, it is likely that, most holders of medical records will continue to send employers an individual's complete medical records (including genetic information).⁸⁴ In addition, such medical records are also likely to contain an individual's family members' information.⁸⁵ Therefore, as science advances and genetic information becomes more prevalent in medical records, there are further risks of discrimination on the basis of genetic information, as well as creating further privacy issues for individuals and family members. Perhaps this is an issue that might be reviewed and considered by the Genetic Nondiscrimination Study Commission.

Criticism can also be directed at the exceptions. As highlighted, the exceptions contained in GINA arguably dilute the effectiveness of the legislation, and increase the accessibility of genetic information.

On evaluating GINA, it is important to look at whether it is succeeding in doing what it set out to do, in particular, alleviating the fear of individuals and reluctance to undergo genetic testing. In a survey carried out in January 2011, it was found that individuals are still worried about the privacy of their genetic information, which is consequently having an adverse effect on their decisions to undergo genetic testing.⁸⁶ Almost three quarters of those surveyed conveyed concern about health insurers accessing their genetic information, and more than half of those surveyed expressed that they were "*extremely concerned*" about such access.⁸⁷ It is questionable therefore whether fears of discrimination have been eliminated, as was the intention of GINA.⁸⁸ The study also highlighted a lack of awareness of GINA and the protections in place for genetic information. In addition, the study indicated that the majority of physicians surveyed were not aware of the legal protections. Further studies carried out in 2012 and 2013 also

⁸² ADA

⁸³ Mark A. Rothstein 'GINA's Beauty is Only Skin Deep' (2009) 22 GeneWatch 2 9, 10

⁸⁴ Mark A. Rothstein and Meghan K. Talbott 'Compelled Disclosure of Health Information: Protecting Against the Greatest Potential Threat to Privacy' (2006) 295 Journal of the American Medical Association 2882-2885

⁸⁵ Mark A. Rothstein, 'GINA's Beauty is Only Skin Deep' (2009) 22 Gene Watch 2 9, 10

⁸⁶ 5th Cogent Genomics, Attitudes and Trends study, conducted by Cogent Research, Cambridge, Massachusetts (January 2011). See also Samuel W. Anderson, 'Genetic Privacy Worries on the Rise' – Interview with Christy White, Cogent Research (2011) 24 Gene Watch 1 15

⁸⁷ Ibid.

⁸⁸ Robert Klitzman, 'Views of Discrimination among Individuals Confronting Genetic Disease' (2010) 19 Journal of Genetic Counseling 68

indicate a lack of awareness of genetic science and the legal protections in place, amongst medical professionals,⁸⁹ consumers⁹⁰ and the general public.⁹¹ These studies clearly indicate the need to incorporate some form of awareness raising educational campaign for the medical community, employers and insurance companies, as well as the public, to inform of the impact of genetics and the legal protections in place.

As highlighted, we can also look to GINA as an example of preemptive federal level legislation, enacted not in response to widespread discrimination and abuse, but in recognition of the speed at which science and technology is advancing and the need for the law to be proactive in its response to these advances. It is observed that well-drafted legislation can “*preempt, or at least mitigate, the perils of genetic discrimination on the horizon.*”⁹² GINA is a preemptive law with the intent of anticipating a potential type of discrimination, in light of rapid advances in genetic science.

In light of the preemptive aspect of GINA, it may be challenging to accurately evaluate its effectiveness. In this regard, Roberts has also commented on the preemptive nature of GINA and states “*we may never know if the statute was hugely successful or completely unnecessary.*”⁹³ Similarly, it is noted that there have been very few cases taken under GINA, so optimistically this could be illustrative of a successful law, actually deferring prohibited conduct. Further, the rapid advances in genetic science in the US (with reference to the falling cost of sequencing the human genome, the increasing popularity of DTC genetic testing, as well as the recently announced Human Brain Project⁹⁴) may also be evidence that GINA is subtly instilling confidence and allowing science to advance.

Overall, the US did well to reconcile the competing interests at stake including concerns of genetic discrimination, genetic privacy and the legitimate interests of employers and insurers. We can certainly look to the US and reflect upon how it has achieved this balance, particularly in light of its own social and historic background.

⁸⁹ Amanda L. Laedtke *et al*, ‘Family Physicians’ Awareness and Knowledge of the Genetic Information Non- Discrimination Act (GINA)’ (2012) 21 Journal of Genetic Counseling 345

⁹⁰ Dawn C. Allain *et al*, ‘Consumer awareness and attitudes about insurance discrimination post enactment of the Genetic Information Non- Discrimination Act (2012) 11 Familial Cancer 4 637

⁹¹ Suzanne B. Haga *et al*, ‘Public knowledge of and attitudes toward genetics and genetic testing’ (2013) 17 Genetic Testing and Molecular Biomarkers 4 327

⁹² Eric Mills Holmes, ‘Solving the Insurance/Genetic Fair/Unfair Discrimination Dilemma in Light of the Human Genome Project’ (1996- 1997) 85 Kentucky Law Journal 503, 647

⁹³ Jessica L. Roberts, ‘Preempting Discrimination: Lessons from the Genetic Information Nondiscrimination Act’ (2010) 63 Vanderbilt Law Review 2 439, 488

⁹⁴ John Markoff, ‘Project Seeks to Build Map of the Human Brain’ *New York Times* (New York, 13 February 2013) http://www.nytimes.com/2013/02/18/science/project-seeks-to-build-map-of-human-brain.html?pagewanted=all&_r=0 (accessed 10 May 2013)

Finally, it is pointed out that GINA is part of the larger health care debate in the US, and is to a certain extent, overshadowed by the ongoing health care reform process. Arguably the bigger issue here is access to adequate health insurance and the societal problems this gives rise to. The necessity to acknowledge and tackle access to health care generally is perhaps more important than addressing the regulation of genetic information.⁹⁵ Although a thorough discussion of health care in the US is outside the scope of this thesis, it is necessary to refer to recent developments in the area of healthcare and highlight the introduction of 'Obamacare'.⁹⁶

3. Australia

The second part of this chapter will highlight the position in Australia and how it has responded to the regulation of genetic information. In particular it will examine the major five- year national project in this area. This project was the first of its kind and revealed the reality of advancing genetic science and the ethical and legal issues which are quickly emerging. Australia is a world leader in this regard, as the project was the first national level investigation into this issue. This section will highlight the aims and objectives of this study and its impact on the Australian framework in this area. This section will also address the work of the Australian Law Reform Commission (ALRC) in this area, which undertook a significant analysis of the key issues arising in this area, culminating in a report entitled *Essentially Yours: The Protection of Human Genetic Information in Australia* (ALRC Report).⁹⁷ The report addressed a range of areas including employment and insurance, and made recommendations on the Australian regulatory position in this regard.

This section will highlight the regulatory approach taken in Australia, for example, the choice to amend existing legislation (as opposed to adopting new stand-alone genetic specific legislation). It will also highlight the choice of a non-discrimination approach as well as a privacy approach and the regulatory recommendations that have been made in this regard. It will focus on the federal level initiatives in Australia.

In terms of a comparative benchmark, it is necessary to mention that healthcare in Australia is universally accessible, placing an emphasis on private insurance, particularly private life insurance. Therefore there is a key difference between Australia and the US in this area. From a comparative perspective, it is also

⁹⁵ Benjamin S. Winford, "The Genetic Information Nondiscrimination Act: Fear Factor or Fantasy Island?" (2008) 38 Hastings Center Report 6 11-12

⁹⁶ Patient Protection and Affordable Care Act of 2010 Pub. L. No. 111- 148, 124 Stat. 119 (2010). For discussion see, Tom Baker, 'Health Insurance, Risk and Responsibility after the Patient Protection and Affordable Care Act' (2011) 159 University of Pennsylvania Law Review 6 1577

⁹⁷ Australian Law Reform Commission, *Essentially Yours: The Protection of Human Genetic Information in Australia* (ALRC 96, 2003) ("ALRC Report")

noted that Australia did not have the same stark history of eugenics motivating genetic based legislation, as the US did. The work and the investigation of the ALRC, as well as the empirical work carried out appears to have been prompted by the increasing accounts of genetic discrimination being reported and investigated, as well as increasing media coverage in the area of genetic discrimination. The work also took place in recognition of the growing awareness of these issues at international level, particularly as regards the position of the UN and the Council of Europe in this area.

3.1 Genetic discrimination in Australia

The following sections will outline the evidence of genetic discrimination in Australia and the empirical research that has been carried out. In the late 1990s and early 2000s discussion and accounts of genetic discrimination were gathering momentum and concern about the implications of genetic advances were becoming increasingly apparent. The first documented study of genetic discrimination in Australia was made in 1998.⁹⁸ In 2000 a further study by Barlow- Stewart and Keays propelled the debate on genetic discrimination in Australia and highlighted the area as one which merited attention.⁹⁹ These studies primarily concerned the areas of life insurance and employment and identified cases of genetic discrimination in these areas. Arguably, these studies were part of the motivation behind the ALRC Report, in terms of verifying the reality of genetic discrimination.¹⁰⁰

3.1.1 The Genetic Discrimination Project

Before detailing the work of the ALRC, this section will highlight the Genetic Discrimination Project (GDP), which provided key research and evidence, and complemented the work of the ALRC in this area. In 2002 the first national study of the nature and extent of genetic discrimination was initiated in Australia.¹⁰¹ This study was funded by the Australian Research Council (for the period 2002 to 2005) and was commissioned in response to the need for comprehensive, empirical research about the incidence of genetic discrimination in an age of advancing genetic science.¹⁰² It built upon the previously documented cases of genetic discrimination in Australia.

⁹⁸ Sandra Taylor, 'A Case Study of Genetic Discrimination: Social Work and Advocacy within a new context' (1998) 51 *Australian Social Work* 51

⁹⁹ Kristine Barlow- Stewart and David Keays, 'Genetic Discrimination in Australia' (2001) 8 *Journal of Law and Medicine* 250. See also David Keays, 'When is Genetic Discrimination Justified?' (2000) 19 *Monash Bioethics Review* 79

¹⁰⁰ Margaret Otlowski *et al*, 'Genetic discrimination: Too few data' (2003) 11 *European Journal of Human Genetics* 1- 2

¹⁰¹ For further details see www.gdproject.org (accessed 25 July 2013)

¹⁰² *Ibid*

This Australian study inquired into the nature and degree of genetic discrimination, from the standpoint of consumers, third parties and the legal system.¹⁰³ By adopting such a multifaceted approach, it ensured that the views of all interested stakeholders were considered. It also acknowledges the presence of competing rights in this area.¹⁰⁴ The aims of this study included the advancement of existing knowledge at a national and international level, the examination of consequences of genetic discrimination in Australia, (from a social and legal perspective), with a view to facilitating recommendations as to the appropriate regulation of the area. Another aim was to gather initial empirical data, to enhance future research endeavours in the area.¹⁰⁵

3.1.2 Methods and findings of the Genetic Discrimination Project

In terms of the methods used in this study, it is observed that the data was gathered by way of interviews, direct dealing and contact with third parties and analysis of documents. The project analysed social and legal aspects of genetic discrimination, particularly in the contexts of employment and insurance. The project highlighted the methodological challenges as regards the investigation of genetic discrimination.¹⁰⁶

It confirmed experiences of genetic discrimination and examined the use of genetic test results in life insurance underwriting in Australia.¹⁰⁷ The study found that there were reported incidents of genetic discrimination, particularly as regards access to life insurance. The results indicated that genetic discrimination by life insurers was most concerning to the respondents in the study.¹⁰⁸

In terms of highlighting the issues arising, the study provided the first empirical evidence of the existence of genetic discrimination in the life-insurance industry, and other areas. It also highlighted the fear of discrimination and found that some individuals were reluctant to engage in genetic testing that could be advantageous for their future health as a result of the fear that the results could act as a barrier to access to life insurance or a family member's access to life

¹⁰³ Sandra Taylor *et al*, 'Investigating Genetic Discrimination in Australia: opportunities and challenges in the early stages' (2004) 23 *New Genetics and Society* 225, 227

¹⁰⁴ Margaret Otłowski *et al*, 'Practices and Attitudes of Australian Employers in relation to the use of genetic information: Report on a National Study' (2009- 2010) 31 *Comparative Labor Law and Policy Journal* 637, 639

¹⁰⁵ <http://www.gdproject.org/whatisit/index.php> (accessed 21 June 2013)

¹⁰⁶ Susan Treloar *et al*, 'Methodological Considerations in the study of genetic discrimination: A review' (2004) 7 *Community Genetics* 161- 168. See also Sandra Taylor *et al*, 'Investigating Genetic Discrimination in Australia: Perceptions and Experiences of Clinical Genetics Service Clients Regarding Coercion to Test, Insurance and Employment' (2007) 5 *Australian Journal of Emerging Technologies and Society* 2 63

¹⁰⁷ Margaret Otłowski *et al*, 'Investigating Genetic Discrimination in the Australian Life Insurance Sector: Use of Genetic Test Results in Underwriting (1999- 2003)' 14 *Journal of Law and Medicine* 367

¹⁰⁸ *Ibid*

insurance.¹⁰⁹ These concerns highlight the need for further research and regulation of this area.

The project also reported genetic discrimination in employment. The employment context was investigated particularly in light of previously reported cases of genetic discrimination in this area.¹¹⁰ The objective of this element of the study was to document employer practices and policies as regards use of genetic information and to highlight the main issues arising for employers in terms of ascertaining an employee's genetic profile.¹¹¹ The study reported incidents of fears of genetic discrimination and highlighted that this could impact upon access to genetic technologies. It also examined potential future interest in and use of genetic information by third parties, for example, employers.¹¹² It found that interest and use in genetic information is likely to increase as genetic technologies become more accessible and available to third parties.¹¹³ The study also noted the potential for genetic information to be gathered from various sources including not only genetic and non- genetic tests, but also clinical observation and inference from family history, thereby highlighting the familial nature of genetic information.¹¹⁴

The project concluded with a number of recommendations. Specifically, it made recommendations in respect of consumers, life insurance, employment and the legal system. These recommendations largely reflect the recommendations of the ALRC Report.¹¹⁵

The in-depth and expansive empirical research carried out in this study was a much needed and welcomed contribution to the debate surrounding advancing genetic science and use of genetic information, as well as the growing body of evidence in this area. Although the study is limited to Australia, it has huge significance in the broader community and reveals valuable insights into the reality of this issue. It provides details as to the particular issues arising in this area, from various perspectives. This project is certainly something that we can

¹⁰⁹ Sandra Taylor *et al*, 'Investigating Genetic Discrimination in Australia: Perceptions and Experiences of Clinical Genetics Service Clients Regarding Coercion to Test, Insurance and Employment' (2007) 5 Australian Journal of Emerging Technologies and Society 2 63

¹¹⁰ Kristine Barlow- Stewart and David Keays, 'Genetic Discrimination in Australia' (2001) 8 Journal of Law and Medicine 250, 250

¹¹¹ Margaret Otlowksi *et al*, 'Practices and Attitudes of Australian Employers in relation to the use of genetic information: Report on a National Study' (2009- 2010) 31 Comparative Labor Law and Policy Journal 637, 639

¹¹² For further discussion, see Margaret Otlowksi *et al*, 'Australian empirical study into genetic discrimination (2002) 4 Genetic Medicine 392. See also Margaret Otlowksi *et al*, 'Major study commencing into genetic discrimination in Australia' (2002) 10 Journal of Law and Medicine 41

¹¹³ Margaret Otlowksi *et al*, 'Practices and Attitudes of Australian Employers in relation to the use of genetic information: Report on a National Study' (2009- 2010) 31 Comparative Labor Law and Policy Journal 637, 690

¹¹⁴ *Ibid* at 638

¹¹⁵ For further details: <http://www.gdproject.org/reports/FINAL%20GDP%20Recommendations.pdf> (accessed 15 July 2013)

learn from in the EU. The research gathered throughout this project validates the initial claims made in respect of genetic discrimination and underscores the work of the ALRC in this area.

3.2 Law Reform Commission Report

From 2001 to 2003, the ALRC carried out an extensive inquiry into misuse of genetic information across a wide range of contexts in Australia. The ALRC Report is the result of a significant, two-year investigation by the ALRC and the Australian Health Ethics Committee of the National Health and Medical Research Council.¹¹⁶

The inquiry was motivated by concerns about privacy and discrimination, especially in the contexts of insurance and employment, and about ethical and other issues arising in scientific research, clinical practice, and the use and collection of genetic databases. There was a growing concern in this area, as reflected by the reported cases of genetic discrimination. The ALRC Report is the result of a comprehensive research and community engagement endeavour. It was described as “*the most comprehensive analysis of the ethical, legal and social consequences of the ‘New Genetics’ ever undertaken.*”¹¹⁷ The Terms of Reference governing the ALRC Report were to make recommendations as regards the most appropriate means of protecting against unfair discrimination, the protection of privacy, as well as acknowledging the need for high ethical standards.¹¹⁸

The Report was drafted against the backdrop of the international position in this area.¹¹⁹ In particular, the Report referred to the UNESCO Universal Declaration on the Human Genome and Human Rights 1997, which highlights, *inter alia*, the need to protect human dignity and prohibit discrimination on the basis of genetic characteristics.¹²⁰ It also referred to the Council of Europe Convention on Human Rights and Biomedicine, Article 11 of which prohibits discrimination on the grounds of genetic heritage.¹²¹ The growing awareness of this issue at international level and the resulting need to address this area informed the recommendations of the ALRC to a certain degree, as did the GDP that was also gathering momentum in Australia at this time.

Although it was found that there was little evidence of Australian employers [currently] using genetic information, it was viewed as inevitable that, as tests become cheaper and more reliable, employers would seek to use this information

¹¹⁶ For details of full report: <http://www.alrc.gov.au/publications/report-96> (accessed 17 April 2013)

¹¹⁷ ALRC Report, Executive Summary

¹¹⁸ ALRC Report, Executive Summary

¹¹⁹ ALRC Report, paras 9.12- 9.16

¹²⁰ ALRC Report, para 9.12

¹²¹ ALRC Report, para 9.15

in the future. The Report acknowledged that as genetic science and technology further advance, it is likely that genetic tests will become more sophisticated and accessible. Therefore it is anticipated that there will inevitably be increased incentives and financial motivation for employers to engage in and seek genetic testing.¹²² The GDP also confirmed this view, with a number of employers expressing interest in the future use of genetic tests if they became more reliable.¹²³ This acknowledgement highlights the support for taking a proactive approach to this issue and recognising the increasing potential for misuse of science by third parties. It also perhaps indicates the need for further, more detailed empirical research to ascertain the nature and prevalence of use of genetic information and genetic tests.

The following sections will look at the particular findings and recommendations of the ALRC Report, and will highlight the regulatory frameworks used. This section will focus on the findings in the areas of employment and insurance.

3.2.1 Choice of regulatory framework

In terms of regulatory models chosen, it is noted that there is not one clear framework used. It is observed that the ALRC viewed this issue both from a disability non-discrimination perspective and also from a privacy perspective, to take account of the misuse of genetic information leading to discrimination and the privacy concerns arising from disclosure and use of genetic information.

The Report provides interesting insights into the concept of genetic information and the appropriate regulatory regime. It considered whether it would be preferable to amend existing anti-discrimination laws to clarify their application to this area, or whether it would be preferable to enact new legislation dealing specifically with genetic discrimination. The Report states that a separate regulatory regime for genetic information is unnecessary.¹²⁴ It rejected the concept of genetic exceptionalism, which was adopted in the US, and which is also a feature of many of the European legislative endeavours in this area.

The Report concluded that discrimination on the grounds of genetic status should continue to be dealt with under the framework of existing federal, state and territorial anti-discrimination laws, subject to the legislative amendments and other safeguards as recommended by the Report.¹²⁵ In coming to this position, the ALRC examined various submissions and consultations. Some submissions to the inquiry advocated in favour of a new regulatory regime. The advantages of such an approach were highlighted, such as *“the advantage of greater certainty*

¹²² ALRC Report, paras 29.33- 29.34

¹²³ Margaret Otlowski *et al*, ‘Practices and Attitudes of Australian Employers in Relation to the Use of Genetic Information: Report on a National Study’ (2009- 2010) 31 Comparative Labor Law and Policy Journal 637, 681

¹²⁴ ALRC Report, para 9.55

¹²⁵ ALRC Report, Recommendation 9-1

of protection, heightened visibility, and the consequential effect of public consciousness raising."¹²⁶

The ALRC also received submissions in support of an approach focused on amending the existing legal framework. Arguments in this respect were based on the rationale that it was *"the most logical, least interventionist approach."*¹²⁷ A stand-alone approach was deemed *"unnecessary and undesirable."*¹²⁸ In this regard, it was also submitted that *"retaining genetic discrimination within [the] conceptual framework of existing anti-discrimination legislation will ensure that we do not afford different levels of protection to people with disabilities diagnosed by genetic testing, or future or imputed disabilities based on predictive genetic testing compared with other people with disabilities."*¹²⁹ In respect of addressing genetic discrimination in employment, the Report looked at the issue from the perspective of existing disability discrimination legislation and considered the amendment of such legislation.

Similarly, the Report found that a stand-alone approach was not the appropriate way forward in respect of protecting the privacy of genetic information. The Report concluded that *"while some inadequacies in the existing legislative privacy framework can be identified, these are best remedied through changes to general information and health privacy laws (including the Privacy Act), rather than through developing a new regulatory framework for the protection of genetic information specifically."*¹³⁰ Therefore, in the privacy context, the ALRC also recommended the amendment of existing legislation.

Having clarified the regulatory approach advocated by the ALRC, the following sections will look at the particular issues and concerns raised in the employment and insurance contexts, and will highlight the main recommendations proposed.

3.3 Recommendations of the Law Reform Commission

The Report includes 144 recommendations for reform, encompassing protection against unfair discrimination in employment and insurance. Although the Report is a wide-ranging analysis of the issues arising in respect of genetic discrimination and genetic privacy across a broad spectrum, for comparative purposes, this section will focus on the contexts of insurance and employment and the key recommendations made. The Report looked primarily at the issues of discrimination and privacy.

¹²⁶ Centre for Law and Genetics, Submission G048 (14 January 2002)

¹²⁷ Genetic Discrimination Project Team, Submission G252, 20 December 2002

¹²⁸ Ibid

¹²⁹ Anti-Discrimination Board of New South Wales, Submission G157, 1 May 2002

¹³⁰ ALRC Report, para 7.10

3.3.1 Genetic information in employment

In Australia, as noted, there is no link between employment and health insurance. Therefore the focus of the Report in this context was on the balance between the interests of employers, employees and the public, and identifying the inappropriate use of genetic information and the ethical and legal concerns this gives rise to. The Report also looks at the regulatory framework in respect of occupational health and safety, which aims to prevent occupational disease. In the employment context, the Report highlighted the current regulatory position. At federal level, the Disability Discrimination Act 1992 is particularly relevant.¹³¹ This law prevents discrimination by an employer against an employee on the grounds of disability.¹³²

In terms of changes to existing legislation, the Report advocated a general prohibition on the use of genetic information by employers.¹³³ It recommended that the Disability Discrimination Act be amended to clarify that an employer is prohibited from requesting or requiring genetic information from an applicant or a current employee, except under certain circumstances.¹³⁴ In this regard, it also recommended that the definition of 'disability' under the Disability Discrimination Act should be amended to clarify that the legislation applies to discrimination based on genetic status.¹³⁵ Although the definition of disability under the Disability Discrimination Act was reasonably broadly drafted and arguably already included discrimination on the grounds of genetic predisposition to disability, the objective was to clarify the inclusion of this ground.

The ALRC Report also recommended the inclusion of a number of limited exceptions, including on occupational health and safety grounds and for the protection of third party safety.¹³⁶ The Report therefore acknowledged the competing interests at stake in this debate. It highlighted the need to ensure the health and safety of employees and it explored the current obligations in this regard, particularly in relation to workplace hazards and managing potential environmental risks.¹³⁷

The awareness of competing rights and the inclusion of exceptions are reflective of the balance reached in the US legislation in the employment context, as discussed. However, the Report recommended that in utilising these exceptions in the employment context, there must be strong evidence of a connection between the working environment and the development of the condition. There must be evidence that the condition may result in serious damage to the health

¹³¹ Disability Discrimination Act 1992 Act No. 135 of 1992 as amended

¹³² Ibid

¹³³ ALRC Report, Recommendation 30-1

¹³⁴ ALRC Report, Recommendation 31-3

¹³⁵ ALRC Report, Recommendation 9-3

¹³⁶ ALRC Report, Recommendations 32-1, 32-2, 32-3, 32-4

¹³⁷ Ibid

or safety of the employee and in addition, the genetic test in question must be a scientifically accurate and reliable method by which to screen for the condition.¹³⁸ There was also a clear recognition of the need to ensure the scientific reliability and accuracy when considering the use of genetic information in employment.

The Report also investigated the privacy issues arising in the employment context.¹³⁹ It was recommended that policies would need to be developed addressing how genetic information is stored and protected against being used for other purposes. In the privacy context, the Report also highlighted the current regulatory position. It found that there was a lack of comprehensive privacy protection under workplace relations legislation.¹⁴⁰ The Report recommended, *inter alia*, the need to amend the Privacy Act,¹⁴¹ “to ensure that employee records are subject to the protection of that Act, to the extent that they contain genetic information.”¹⁴² This recommendation acknowledges the right to privacy in the information in question and the other personal rights in genetic information, such as the right to autonomy and dignity of the person.

3.3.2 Genetic information in insurance

Consideration of the position in Australia must be viewed in light of the health care system. In Australia, there is a national health system and private health insurance is not risk-rated. Therefore concerns regarding the potential for genetic discrimination are directed primarily at the life insurance industry.¹⁴³ Indeed, this is reflected in the GDP which found that genetic discrimination is most worrying in the life insurance industry.¹⁴⁴ Life insurance therefore was the main context within which genetic discrimination occurred in the Australian investigation, but this does not eliminate the potential of discrimination in health or other insurance contexts in other countries.

In the insurance context, the ALRC Report focuses on two particular areas. First, it looks at what genetic information is collected by insurance companies, the manner in which it is used for underwriting purposes, and the issues that arise in this regard, including potential unlawful discrimination.¹⁴⁵ Secondly, the Report addresses the privacy issues that might arise in the insurance context. It looks at

¹³⁸ ALRC Report, Recommendations 32-1, 32-4

¹³⁹ ALRC Report, chapter 34

¹⁴⁰ ALRC Report, para 34.19. See Workplace Relations Act 1996 Act No. 86 of 1988, as amended

¹⁴¹ Privacy Act 1988 Act No. 119 of 1988, as amended

¹⁴² ALRC Report, Recommendation 34- 1

¹⁴³ Sandra D. Taylor *et al*, ‘Investigating Genetic Discrimination in Australia: opportunities and challenges in the early stages’ (2004) 23 New Genetics and Society 2 225, 226

¹⁴⁴ Sandra Taylor *et al*, ‘Investigating Genetic Discrimination in Australia: Perceptions and Experiences of Clinical Genetics Service Clients Regarding Coercion to Test, Insurance and Employment’ (2007) 5 Australian Journal of Emerging Technologies and Society 2 63, 70- 72

¹⁴⁵ ALRC Report, chapters 25 and 26

whether and to what extent, a regulatory framework is needed to protect the privacy of genetic information in insurance (and other contexts).¹⁴⁶

On examining the non-discrimination regulatory framework in the insurance industry, it observed that the area is governed by legislation and common law principles, as well as soft law standards in the industry.¹⁴⁷ In terms of non-discrimination law at federal level, several pieces of legislation are relevant, including the Racial Discrimination Act 1975, the Sex Discrimination Act 1984 and the Disability Discrimination Act 1992. In addressing the use of genetic information in insurance in Australia, it is particularly relevant to refer to the Disability Discrimination Act 1992.¹⁴⁸

This Disability Discrimination Act makes it unlawful to discriminate in the provision of goods and services, and 'services' include insurance services. However, the Act recognises the necessity to engage in risk classification and to distinguish between insurance applicants on the grounds of health profile, (as well as family history of illness).¹⁴⁹ Accordingly, it is permissible for life insurers to effectively discriminate between individuals on the grounds of their estimated future health risks, for underwriting purposes. The Act provides that if insurers base their actions in clear actuarial or valid statistical data, they will not be in violation of the legislation. The Report also made recommendations in support of these insurance provisions.¹⁵⁰ It recognised the fundamental principles of insurance law including the requirement of mutual disclosure and the doctrine of utmost good faith.¹⁵¹

Section 27 of the ALRC Report specifically discusses the issue of scientific reliability and actuarial relevance.¹⁵² This is a key point in this discussion and an issue that should always be taken into account when considering use of genetic information. Use of scientifically unreliable or inaccurate information may exacerbate the potential for misuse of the information by third parties. This is relevant both in the employment and insurance contexts.

In terms of the specific issue of use of genetic information by life insurers, it is pointed out that there is no firm legislative position. Instead, there has been a voluntary, cooperative approach taken by the insurance industry, as was recognised in the Report.¹⁵³ In the context of insurance and genetic information, it is therefore noted that it has been primarily a soft law approach in place, as opposed to a legislative approach. In 2000 a voluntary policy was introduced

¹⁴⁶ ALRC Report, chapter 28

¹⁴⁷ ALRC Report, paras 26.6 and 26.7

¹⁴⁸ Disability Discrimination Act 1992 Act No. 135 of 1992 as amended

¹⁴⁹ Ibid

¹⁵⁰ ALRC Report, Recommendation 26-1,

¹⁵¹ Ibid

¹⁵² ALRC Report, Recommendation 27-1

¹⁵³ ALRC Report, para 26.7

which provided that no insurer shall require that an applicant or insured person undergo genetic testing. However, pursuant to this policy, insurance applicants must disclose genetic test results previously taken, or if they are aware of family members' genetic test results, then they must disclose these details in circumstances where they are applying for a new policy or amending an existing policy.¹⁵⁴

In contrast to the employment context, the Report did not make any concrete recommendations as regards legislative amendments.¹⁵⁵ In terms of recommendations, the Report found that there should be no departure from the fundamental principle underlying the industry, that is, equality of information between the insurance applicant and the insurance company.¹⁵⁶ The Report did however make a number of recommendations with a view to ensuring that the inappropriate use of genetic information does not take place in the insurance industry. These recommendations include ensuring the assessment of the scientific reliability and the actuarial relevance of the genetic test in question if use is intended in the insurance context,¹⁵⁷ the ongoing education of insurance agents in the industry, with a view to avoiding the inappropriate discriminatory use of genetic information,¹⁵⁸ and the introduction of clarity as regards a complaints mechanism in circumstances where an inappropriate decision has been made.¹⁵⁹

The Report also recommended that the Human Genetics Commission should maintain a watching brief to monitor developments in the insurance industry as regards the use of genetic information, at national level and on an international level, with the objective of amending Australian insurance practices as necessary.¹⁶⁰

As noted, the Report also considered the privacy issues that might arise in the insurance context, and it looks at the effectiveness of the current regulatory system. It notes that the collection, use, storage and disclosure of an insurance applicant or an insured's personal information by insurance companies is regulated by the Privacy Act 1988.¹⁶¹ The Report found that there were no significant concerns identified and concluded that the basic privacy protection framework in the insurance context is adequate.¹⁶² The Report did however highlight a number of concerns in this respect, including the quality of consent to collection and use of genetic information by insurance companies, the collection

¹⁵⁴ Investment and Financial Services Association, IFSA Standard 11.00 'Genetic Testing Policy' (2002), IFSA

¹⁵⁵ ALRC Report, part G Recommendations

¹⁵⁶ ALRC Report, Recommendation 26-1

¹⁵⁷ ALRC Report, Recommendation 27-1

¹⁵⁸ ALRC Report, Recommendation 27-10

¹⁵⁹ ALRC Report, Recommendation 27-9

¹⁶⁰ ALRC Report, Recommendation 26-2

¹⁶¹ Privacy Act 1988 Act No. 119 of 1988, as amended

¹⁶² ALRC Report, para 28.19

of family medical history by insurance companies and the sharing of information between certain organisations.¹⁶³

3.4 The Human Genetics Advisory Committee

An important recommendation in the ALRC Report is for an expert body to be established to provide advice to Australian governments about current and emerging issues in human genetics.¹⁶⁴ An independent, expert advisory body, the Human Genetics Advisory Committee was established in 2005, and is working on implementing the recommendations of the ALRC Report. The Committee will also provide continuing advice to government on technical and strategic issues in human genetics.¹⁶⁵ The merits and potential value of this endeavour are recognised.

It is anticipated that this Committee will respond to the growing ethical and legal challenges and recognise advances in genetic science. Indeed this Committee is similar to the proposed Genetic Nondiscrimination Study Commission in the US, in terms of role and objectives. There is a clear need for such a body in this area, and the EU could certainly learn from such endeavours.

3.5 Government response to the recommendations and implementation

The Australian Government issued an official response to the recommendations of the ALRC Report on 9 December 2005, and accepted many of the recommendations of the report.¹⁶⁶ These recommendations are currently being implemented. For example, the definition of disability under the Disability Discrimination Act has been amended to explicitly include genetic predisposition to a disability, thereby broadening the scope and application of the legislation.¹⁶⁷ There has also been the introduction of a provision, which prohibits employers from requesting or requiring genetic information from a job applicant or employee except where reasonably required for purposes not involving genetic discrimination.¹⁶⁸

In the privacy context, the Report had recommended the expansion of the definition of health information to explicitly include “*genetic information*.” In this regard, the Privacy Act 1988 has been amended by the Privacy Legislation Amendment Act 2006, which inserted a new subsection to include “*genetic*

¹⁶³ ALRC Report, Recommendations 27-1, 28-1, 28-2, 28-3, (paras 28.67, 28.68, 28.69)

¹⁶⁴ ALRC Report, Recommendation 5-1

¹⁶⁵ <http://www.nhmrc.gov.au/about/committees-nhmrc/human-genetics-advisory-committee-hgac> (accessed 25 May 2013)

¹⁶⁶ See <http://www.alrc.gov.au/inquiries/health-and-genetics/full-australian-government-response-alrc-report-96> (accessed 20 March 2013)

¹⁶⁷ Disability Discrimination Act 1992 Act No. 135 of 1992 as amended

¹⁶⁸ Disability Discrimination and other Human Rights Legislation Amendment Act 2009 Act No. 70 of 2009, schedule 2, s 20, ss 3(b)

information.” It states “genetic information about an individual in a form that is, or could be, predictive of the health of the individual or a genetic relative of the individual.”¹⁶⁹ The Government support for these recommendations is significant in lending credibility to these issues and further raising awareness in this area.

3.6 Evaluation of the position in Australia

The Australian position offers an interesting perspective to this debate. The GDP provides an interdisciplinary and practical insight into the reality of genetic discrimination and other misuses of genetic information. This national level study gathered key empirical evidence of the scenarios and instances in which such discrimination and other misuse can occur. The in-depth research and public engagement in this area contributed greatly towards the understanding of this area, not only in Australia, but also worldwide. We can look to this project as evidence of the incidence of genetic discrimination and the main issues that arise as regards the use of genetic information. We can also look to this project as a useful model when considering the need for empirical evidence of genetic discrimination in the EU.

The ALRC Report made a significant contribution to the debate in this area in recognising the key ethical and legal issues and formulating recommendations for reform. The establishment of the Human Genetics Advisory Committee is also an admirable step in the right direction and an acknowledgement of the need to keep up to date with advances in science and new challenges that arise. As noted, the EU might learn from this. The Australian Government has accepted the majority of the recommendations and implementation is proceeding.

4. Conclusion

From a comparative perspective, there is a growing recognition of the need to respond to scientific and technological advances, as well as a need to consider appropriate legislative and policy intervention. By way of comparative benchmarks in this thesis, this chapter highlighted the response to advancing genetic science in the US and Australia, both of which provided different lenses through which to view this issue. It is observed that there are both similarities and differences that can be identified with the various approaches. While the US took primarily a non-discrimination approach and recognised the merits of pursuing a new regulatory regime, specifically tailored to the issue of genetic discrimination, Australia rejected the option of a new regulatory regime and viewed the issue from both a non-discrimination and a privacy perspective. In terms of chosen regulatory frameworks, the predominant approach that can be identified is therefore non-discrimination.

¹⁶⁹ Privacy Legislation Amendment Act No. 99 of 2006. See Schedule 2, 3 ss 6(1)

Although the historical and legal backgrounds of these jurisdictions are different to the EU, it is acknowledged that they are all dealing with similar issues with the advent of this genetic age. As illustrated in part 1 of this chapter, the US is arguably a pioneer in this area. By adopting an anti-discrimination framework, it has introduced federal level legislation, specifically tailored to the regulation of genetic information. Although not a perfect response, it highlights the balance struck in the US.

It is important to note the forward-thinking approach taken by the US, and recognise the value of reacting to advancing technology at an early stage. The merits of this preemptive model must be recognised in acknowledging the importance of keeping up to date with science and technology and anticipating the ethical and legal issues that might arise. Such an approach also operates to enhance public trust and confidence in science, to a certain degree, which ultimately benefits public health, as well as facilitates further scientific innovation and growth.

The US attempted to strike a balance, which is evident in the inclusion of a number of exceptions, recognising the different contours to the debate and acknowledging competing interests. The US also attempted to achieve a degree of flexibility and foresight with the provision of a Genetic Non Discrimination Commission. The shortcomings with the US legislation also provide valuable reference points, particularly in relation to the limited scope of the protection provided and the practical challenges that are likely to be encountered. On evaluation of the position in the US, the broader health care debate is inescapable, and it is submitted that, to a certain degree, it is this debate which has taken primacy over the issue of genetic discrimination.

In the US context, it is also important to note the studies carried out post- GINA, which indicate a lack of awareness of genetic science and the legal protections in place, amongst the general public and the medical profession. This clearly points to the need for some form of awareness raising campaign, and we can certainly take this on board in the EU context.

Australia has also been a leader in this area. The ALRC's Report was a welcomed contribution to this debate and is informative for comparative purposes. It has had a firm impact in this area and the recommendations are currently being implemented. The EU can certainly learn from developments both of these jurisdictions. In particular, it can take note of the major empirical study which took place in Australia and perhaps consider a similar model in this area. The merits of an independent body, similar to the Human Genetics Advisory Committee and the Genetic Nondiscrimination Study Commission, are also recognised, and may perhaps be considered in the EU alongside any legislative proposals.

Having examined the regulatory options and frameworks, and having provided a comparative analysis of the position in the US and Australia, part 3 of this thesis will look at the European perspective in this area, both from the perspective of the Council of Europe and the EU.

PART 3: THE EUROPEAN PERSPECTIVE

Part 2 of this thesis introduced the moral imperative for regulation in this area, and explored the choice of regulatory frameworks that best suit the regulation of genetic information, with a focus on the non- discrimination and privacy models. In addition, it examined the international human rights framework, documenting the evolution in the interpretation and orientation of human rights. It also examined the comparative law benchmarks in this area, highlighting the regulatory choices taken in these comparative jurisdictions. Part 3 of this thesis includes two chapters and looks at the European perspective, encompassing discussion of the Council of Europe (CoE) in providing a moral framework from which to examine this area (chapter 8). Chapter 9 focuses on the EU and will look at the application of current EU law to genetic information and will examine the reach of current non- discrimination and data protection laws.

Chapter 8: Human rights norms in the Council of Europe and the regulation of genetic information

1. Introduction

This chapter will examine the CoE's human rights standards and the relevant instruments and jurisprudence, which shape this position. This chapter will give a chronological overview of the various instruments and documents of the CoE. It will examine the position in response to emerging genetic technology, in providing a moral and ethical framework within which to approach this area. As highlighted, genetic science is advancing at a rapid pace and giving rise to a myriad of ethical and legal dilemmas, with questions arising as to the regulation of such issues. This chapter will explore the relevant instruments, and how they might shape our consideration of the issues of genetic discrimination and genetic privacy from a human rights perspective.

The CoE operates against a basic human rights background, and recognises core principles of public morality.¹ Against this background, many of the conventions address fundamental human rights issues. Firstly, this chapter will look at the ECHR,² and its influence on this area, in emphasising the key position of human rights in the Member States. Of particular relevance is the Convention on Human Rights and Biomedicine, and the subsequent protocol on genetic testing for health purposes. This is an important instrument in providing guidance on how to address these bioethical issues, as well as highlighting the key ethical and legal issues arising. This chapter will also look at other relevant instruments such as the European Social Charter and the Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data. Finally, it will highlight recent policy developments in the area of bioethics, specifically the focus on genetic testing and insurance.

2. European Convention on Human Rights and case law

This section will address the significance of the ECHR, as a key source of fundamental human rights in Europe. It will look specifically at the application of

¹ Gerard Quinn, 'The European Union and the Council of Europe on the Issue of Human Rights: Twins Separated' (2000- 2001) 46 McGill Law Journal 849, 856. See also Krzysztof Drzewicki, 'European Systems for the Promotion and Protection of Human Rights' in Catarina Krause and Martin Scheinin (eds) *International Protection of Human Rights: A Textbook* (2nd edn, Abo Akademi University Institute for Human Rights 2012)

² Convention for the Protection of Human Rights and Fundamental Freedoms ETS 5; 213 UNTS 221 ("ECHR")

the ECHR to the issues of genetic discrimination and genetic privacy. This section will also highlight the connection between EU law and the ECHR.

The ECHR has the objective of protecting human rights and fundamental freedoms. On 4 November 1950 the text of the ECHR was signed and it entered into force on 3 September 1953. The ECHR is recognised as a novel example of a comprehensive human rights framework, with a high profile position on the international stage.³ The ECHR also established the European Court of Human Rights (ECtHR),⁴ which holds responsibility for determining the nature and scope of the rights enshrined in the ECHR. The ECHR provides a key framework for fundamental rights and provides for a number of fundamental rights and freedoms, including, *inter alia*, the right to life, the right to liberty, freedom of expression and freedom of assembly and association. Quinn opines that it was clear that the “*intention of the framers was to protect certain core rights that honoured individual dignity and autonomy.*”⁵

The following section will provide an overview of the overall tenor of the ECHR, with a particular focus on the provisions relating to privacy and non-discrimination. In the context of this debate, of initial importance are Article 8 and Article 14. Article 8 contains four particular rights: the right to respect for private life, for family life, for correspondence and for the home.⁶ Article 8 offers expansive protection and states, *inter alia*, “*Everyone has the right to respect for his private and family life, his home and his correspondences...*”⁷ Therefore, it has been recognised that “*the concept of ‘private life’ covers the physical and psychological integrity of a person; it embraces aspects of an individual’s physical and social identity.*”⁸ It has been noted that elements such as gender identification, name and sexual orientation and sexual life fall within the personal sphere protected by Article 8.⁹ The right to private life, as contained in Article 8 encompasses the concept of personal autonomy, as was found in the case of *Pretty v UK*.¹⁰

In the context of ‘informational privacy’ a key case is *S. and Marper v the United Kingdom*.¹¹ This case concerned whether the retention of DNA and fingerprints

³ Rick Lawson, ‘The European Convention on Human Rights’ in Catarina Krause and Martin Scheinin (eds) *International Protection of Human Rights: A Textbook* (2nd edn, Abo Akademi University Institute for Human Rights 2012) 423. See also Peter van Dijk *et al* (eds) *Theory and Practice of the European Convention on Human Rights* (4th edn, Intersentia 2006)

⁴ ECHR, Article 19

⁵ Gerard Quinn, ‘The European Union and the Council of Europe on the Issue of Human Rights: Twins Separated’ (2000- 2001) 46 McGill Law Journal 849, 857

⁶ *A, B & C v Ireland* (No. 6833/74) judgment of 16 December 2010. In this case, the court referred to a right to personal autonomy in the context of Article 8

⁷ ECHR, Article 8

⁸ Rick Lawson, ‘The European Convention on Human Rights’, in Catarina Krause and Martin Scheinin (eds) *International Protection of Human Rights: A Textbook* (Gummerus Printing 2009) 405

⁹ *Ibid* at 405

¹⁰ *Pretty v the United Kingdom* (No. 3246/02) [2002] ECHR 29 April 2002

¹¹ *S. and Marper v. the United Kingdom* (No. 30562/04) [2008] ECHR 1581 4 December 2008

from innocent individuals is in accordance with human rights law. In this case, the Court held that holding DNA samples of individuals who are arrested but have the charges against them dropped or are later acquitted, is a violation of the right to privacy under the ECHR. This was an aspect of what was referred to as “*informational privacy and there could be little, if anything, more private to the individual than the knowledge of his genetic make-up.*”¹² Article 8 is therefore particularly relevant in the context of the right to privacy and protection of personal information. Further, and importantly in this debate, Article 8 is also relevant in terms of protecting genetic data, as a form of personal information. It is therefore arguable that these protections may also extend to encompass the issue of genetic privacy. This is a seminal case in recognising the right to privacy as regards one’s genetic data. From a human rights perspective, the ECtHR is aware of the issue of personal privacy as a core and fundamental human right. The privacy provisions are reflective of those contained in the UN Declaration on Human Rights, and the ECHR places the right to privacy on a firm international footing.¹³

In addressing the issue of non- discrimination, Article 14 is key and merits discussion. Article 14 prohibits discrimination and contains a non- exhaustive list of covered grounds.¹⁴ Article 14 states “*The enjoyment of the rights and freedoms set forth in this Convention shall be secured without discrimination on any ground such as sex, race... or other status.*”¹⁵ However, it is noted that Article 14 cannot be relied upon on its own and it does not provide a free-standing right not to be discriminated against. It only operates with the other provisions of the Convention. It has been aptly described as having an ‘accessory nature’¹⁶ and ‘no independent existence.’¹⁷ Therefore, any claims brought pursuant to Article 14 must relate to other substantive rights and provisions of the ECHR, for example, the right to privacy. In relation to the non-exhaustive nature of Article 14, Hendriks has referred to the ECHR as “*a living instrument that must be interpreted in present- day conditions.*”¹⁸ This has been particularly reflected in the case law of the ECtHR, which will be discussed next.

¹² Ibid at para 25

¹³ Mark Taylor, *Genetic Data and the Law – A Critical Perspective on Privacy Protection* (Cambridge 2012) 67

¹⁴ For discussion, see Janneke Gerards, ‘The Discrimination Grounds of Article 14 of the European Convention on Human Rights’ (2013) 13 *Human Rights Law Review* 1 99

¹⁵ ECHR, Article 14

¹⁶ Gomien, Harris and Zwaak, ‘Law and Practice of the European Convention on Human Rights and the European Social Charter’ (Council of Europe, 1996) at 346

¹⁷ See for example, the *Belgian Linguistics Case* (1979- 1980) 1 EHRR 578, para 9. See also *Airey v Ireland* (1979- 1980) 2 EHRR 305, para 30

¹⁸ Aart Hendriks, ‘Protection against Genetic Discrimination and the Biomedicine Convention’ (2005) in Jan K.M. Gevers, Ewoud H. Hondius and Joep H. Hubben (eds) *Health Law, Human Rights and the Biomedicine Convention* (Koninklyke Brill N.V. 2005) 212. See also Janneke Gerards, ‘The Discrimination Grounds of Article 14 of the European Convention on Human Rights’ (2013) 13 *Human Rights Law Review* 1 99, 116

Although disability (or ‘genetic information’) is not mentioned specifically as a covered ground, the non-exhaustive list element of this provision would seem to stretch the ambit of protection to cover disability. However, before the case of *Glor v Switzerland*, the ECtHR had not yet found a violation of the right to non-discrimination on the basis of disability.¹⁹ In this case, the ECtHR held that diabetes constituted a disability. The ECtHR reprimanded Swiss authorities for failing to provide reasonable accommodation to Mr Glor by finding a solution which responded to his individual circumstances. It is also noteworthy to point out that the judgment referred to the CRPD,²⁰ which it held “*signalled the existence of a European and worldwide acknowledgement of the need to protect persons with disabilities from discriminatory treatment*” (even though Switzerland had not signed the CRPD).²¹ This case is of great relevance, firstly in placing an emphasis on the CRPD, also in finding a violation on the basis of disability, and indeed by taking a relatively expansive approach to the concept of disability, in line with the CRPD.

Following the decision in *Glor v Switzerland* in which the ECtHR found that discrimination on the grounds of disability is provided for under Article 14, the court made another decision that clarifies and further expands the scope of Article 14. In the case of *G.N. and others v Italy*, the ECtHR made a finding of discrimination on the basis of genetic disease, under Article 14 ECHR.²² The court asserted that even though Article 14 does not mention genetic characteristics, disability or health under its enumerated grounds, the list is not exhaustive.²³ Importantly, the court also found that discrimination on the basis of a genetic disorder is closely linked to disability discrimination. It held that different treatment on the basis of genetic disease does fall within Article 14 of the ECHR. It is noted that the court also referred to Article 21 of the EU Charter of Fundamental Rights.²⁴

Another instructive case of the ECtHR is *Kiyutin v Russia*.²⁵ In this case, the applicant’s application for a residence permit in Russia was refused on the basis that he had tested HIV- positive. The Court referred to the “*blanket and indiscriminate nature*” of the measure in question.²⁶ In this case, evidence indicated that the domestic authorities had rejected his application solely by reference to the statutory provisions without taking into account “*his state of health or his family ties in Russia*.”²⁷ The ECtHR ultimately upheld the claim of discrimination on the grounds of the individual’s health status as HIV positive. It

¹⁹ *Glor v Switzerland* (No. 13444/04) [2009] ECHR 13 April 2009

²⁰ G. A. Res 61/106 (“CRPD”)

²¹ Jarlath Clifford, ‘The UN Disability Convention and its Impact on European Equality Law’ (2010) 6 The Equality Rights Review 11 20

²² *G.N. and others v Italy* (No. 43134/05) [2009] ECHR 1 December 2009

²³ *Ibid*

²⁴ *Ibid*

²⁵ *Kiyutin v Russia* (No. 2700/10) [2011] ECHR judgment of 10 March 2011

²⁶ *Ibid*, para 72

²⁷ *Ibid*, para 73

held that there had been a violation of Article 14 of the ECHR, in conjunction with Article 8.

Discussion of Article 14 necessitates reference to Protocol 12 to the European Convention on Human Rights,²⁸ the objective of which was to expand the scope and field of application of Article 14 beyond the rights included in the ECHR. This protocol can be viewed as a complement to Article 14 and acts to enhance the effectiveness of the non- discrimination provision.

On examining the principle of non- discrimination under the ECHR, it must also be pointed out that the concept of indirect discrimination is not explicitly provided for in the ECHR. However, this concept has been acknowledged by the ECtHR, in *Hugh Jordan v the United Kingdom*,²⁹ and *DH and Others v the Czech Republic*.³⁰ These cases further enhance the non- discrimination protections under the ECHR.

2.1 Evaluation of the ECHR human rights standards

The ECHR is a seminal legal instrument in the area of human rights and has propelled the importance of fundamental human rights amongst the Member States. The ECHR provides a core human rights framework from which to view this area and identify the key issues. It also highlights both a non- discrimination and a privacy approach to protecting human rights that can potentially be applied to the regulation of genetic information. Both the non- discrimination provisions and the privacy provisions are strong and potentially stretch across a wide spectrum, as is particularly reflected in the case law of the ECtHR. Indeed the case law of the ECtHR is reflective of evolving human rights norms in this area, including an interpretation of discrimination on the grounds of disability, and the recognition of genetic privacy.

It is noted that there is a close connection between the EU human rights framework and the ECHR, as will be further highlighted in the next chapter. EU human rights law is largely in line with the ECHR, and this is particularly reflective in the Charter of Fundamental Rights of the EU. The impending future accession of the EU to the ECHR will further enhance this connection and promote the ECHR's significance in EU human rights discourse.³¹ It will narrow the gap in human rights protections and improve the consistency between the various human rights systems.

²⁸ Protocol No. 12 to the ECHR signed on 4 November 2000 and entered into force on 1 April 2005 CETS No. 177

²⁹ *Hugh Jordan v the United Kingdom* (No. 24746/94) [2001] ECHR 4 May 2001

³⁰ *DH and Others v the Czech Republic* (No. 57325/00) [2007] ECHR Chamber judgment 7 February 2006 and Grand Chamber judgment 13 November 2007

³¹ Treaty on the European Union, Article 6(2)

3. European Social Charter

This section will provide a brief overview of the European Social Charter. As a complement to the ECHR (although largely overshadowed by the ECHR³²), the European Social Charter was adopted in 1961 and revised in 1996.³³ The aim of the Charter is to promote and enhance economic activity and social progression for all individuals, and these aims are reflected in the provisions of the Charter. There is a certain emphasis on equality and non- discrimination, and the guarantee of basic human rights and freedoms. It also contains provisions in relation to the protection of health and access to health care. In addition, the Charter contains a right to job training for persons with disabilities as well as special measures in order to assist with obtaining employment.

The Revised European Social Charter adds a right of persons with disabilities to independence, social integration and participation in the life of the community.³⁴ Further, Article E provides for a general non- discrimination clause, including an 'other status' ground, which is reflective of Article 14 of the ECHR. This is an accessory provision, linked to the enjoyment of rights provided for in the Charter.³⁵ The European Social Committee has clarified that disability is a prohibited ground under Article E, although not specifically mentioned in the Article.³⁶ It also does not specifically mention genetic data or genetic status as a covered ground, although it is arguable that such grounds could come within the scope of the 'other status' grounds.³⁷ The ESC is an important instrument of the Council of Europe and a key source of economic and social rights. It is reflective of the progression of human rights within the CoE constitution.

4. Convention for the Protection of individuals with regard to Automatic Processing of Personal Data

The Convention for the Protection of individuals with regard to Automatic Processing of Personal Data³⁸ ("Personal Data Convention") also merits

³² Olivier de Shutter, 'The European Social Charter' in Catarina Krause and Martin Scheinin (eds) *International Protection of Human Rights: A Textbook* (2nd edn, Abo Akademi University Institute for Human Rights 2012) 463

³³ Council of Europe, European Social Charter (Revised) ETS No. 163 ("ESC")

³⁴ ESC, Article 15

³⁵ ESC, Article E

³⁶ *Association Internationale Autisme- Europe (AIAE) v France*, Complaint No. 13/2000, decision on the merits of 4 November 2003, para 51

³⁷ Aalt- Willem Heringa, 'International and Legal Instruments', in Janneke H. Gerards, Aart W. Heringa and Heleen L. Janssen (eds) *Genetic Discrimination and Genetic Privacy in a Comparative Perspective* (Intersentia 2005) 37

³⁸ Convention for the Protection of individuals with regard to Automatic Processing of Personal Data (1981, ETS No. 108)

discussion. Its main relevance lies in the fact that it is still the only binding legal instrument at an international level in the area of data protection, and it potentially has a scope of application on a worldwide scale.³⁹ It concentrates on the significance of the data protection tool as a means of protecting personal privacy.⁴⁰ This Convention takes account of increasing flows of personal data across borders. It is broadly drafted and contains general principles in relation to the protection of personal data and upholding privacy. In this regard, it refers to the concept of “*data quality*”, which sets out a number of provisions in respect of the fair processing of information.⁴¹

There is a close connection between the CoE norms on data protection and the EU’s framework on data protection.⁴² The concept of privacy and data protection is well developed and established in both legal systems, and both contain similar fundamental core principles as regards the collection and use of personal data. Indeed the provisions of this Convention are reflected in the data protection legislative framework in the EU.⁴³ The presence and strength of the Personal Data Convention highlights the overall commitment to the protection of personal data by the Council of Europe. It is further noted that an Additional Protocol to the Personal Data Convention was introduced in 2001, which reinforced the protections in this area.⁴⁴

5. Council of Europe’s Convention on Human Rights and Biomedicine

Since the 1980s, the CoE has been active in the field of bioethics, and has recognised the need to acknowledge that advancing genetic technology gives rise to a corresponding potential for misuse and breach of fundamental rights. These rapid scientific and technological developments have provoked the CoE into action in this area, by developing legal standards with the intention of recognising an individual’s fundamental rights.⁴⁵

³⁹ Maeve Mc Donagh, ‘Balancing Disclosure of Information and the Right to Respect of Private Life in Europe’ (2012) *Journal of Internet Law* 3, 5. For further details see:

http://www.coe.int/t/dghl/standardsetting/DataProtection/default_en.asp

⁴⁰ Fred H. Cate, ‘The EU Data Protection Directive, Information Privacy and the Public Interest’ (1995) 80 *Iowa Law Review* 431, 432

⁴¹ Paul M. Schwartz, ‘European Data Protection Law and Restrictions on International Data Flow’ (1994) 80 *Iowa Law Review* 471, 477

⁴² For further discussion see Marc Rotenberg and David Jacobs, ‘Updating the law of information privacy: the new framework of the European Union’ (2013) 36 *Harvard Journal of Law and Policy* 2 605, 615

⁴³ Council and Parliament Directive 95/46/EC of 24 October 1995 on the protection of individuals with regard to the processing of personal data and on the free movement of such data [1995] OJ L 281/31

⁴⁴ Additional Protocol to the Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data regarding supervisory authorities and cross- border data flows (2001 ETS No. 181)

⁴⁵ Laurence Lwoff, ‘Council of Europe adopts protocol on genetic testing for health purposes’ (2009) 17 *European Journal of Human Genetics* 1374 1-4

The Convention on Human Rights and Biomedicine (Biomedicine Convention) was adopted on 19 November 1996 and entered into force on 1 December 1999.⁴⁶ As of 30 January 2013, 29 countries have ratified this Convention. The Convention focuses on the protection of human dignity, identity and integrity in medicine and it formulates fundamental principles applicable to science and new technologies. The connection between this Convention and the ECHR is acknowledged. It has been observed that the Biomedicine Convention was initiated by connecting bioethics with the human rights approach of the ECHR “*in an attempt to tie the inherent value of the human being into the bioethics debate.*”⁴⁷ In the context of this debate, the Convention sets out a number of principles with regard to genetics, particularly genetic testing, offering guidance on good practice, and acting as a moral framework.

5.1 Overview - The Convention on Human Rights and Biomedicine

This section will give a further overview of the background and objectives of the Biomedicine Convention. The Convention originates from the necessity for “*harmonisation of standards for protection of the individual in the context of scientific and technological developments in medicine and health care.*”⁴⁸ The main aim of the Convention is to facilitate a comprehensive and uniform legal framework to recognise and protect human rights in light of advancing science and technology. It is the first international treaty specifically addressing biomedicine, and has been heralded as “*an epoch- making document opening a new era in bioethics and biolaw.*”⁴⁹ The Convention highlights the need to create uniform standards to address biomedicine in this area. Its broad scope covers all fields involving medical research, and it seeks to regulate the basis of medical care provided by physicians and other healthcare professionals as well as researchers and other stakeholders.

It is noted that the concept of human dignity is evidently “*the bedrock of the Oviedo Convention.*”⁵⁰ Pursuant to the Explanatory Report, “*the concept of human dignity ... constitutes the essential value to be upheld. It is at the basis of most of the values emphasised in the Convention.*”⁵¹ It is pointed out that the Preamble emphasises the importance of human dignity and refers to the principle

⁴⁶ Council of Europe, Convention for the Protection of Human Rights and the Dignity of the Human Being with Regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine, ETS. No. 164 (1997) (“Biomedicine Convention”)

⁴⁷ Eibe Reidel, ‘Global Responsibilities and Bioethics: Reflections on the Council of Europe Bioethics Convention’ (1997) 5 Indiana Journal of Global Legal Studies 1 179, 182

⁴⁸ Henriette Roscam Abbing ‘The Convention on Human Rights and Biomedicine – An Appraisal of the Council of Europe Convention’ (1998) 5 European Journal of Health Law 377, 377

⁴⁹ Maurizio Mori and Demetrio Neri, ‘Perils and Deficiencies of the European Convention on Human Rights and Biomedicine’ (2001) 26 Journal of Medicine and Philosophy 3 323, 323

⁵⁰ Roberto Andorno, ‘The Oviedo Convention: A European Legal Framework at the Intersection of Human Rights and Health’ (2005) 2 Journal of International Biotechnology Law 133, 137

⁵¹ Explanatory Report to the Convention on Human Rights and Biomedicine, para 9

several times.⁵² The emphasis on human dignity as a core concept underlying the Convention reinforces the acknowledgment of human rights as a paramount consideration in bioethics, as well as recognising the primacy of the person. Article 1 includes the guarantee of respect for the individual's integrity and other rights and fundamental freedoms and prohibits any form of discrimination, thereby highlighting dignity and integrity again.⁵³ This emphasis on human dignity is also reflective of some of the UN soft law instruments, discussed in chapter 6.

Further, Article 2 on the primacy of the human being states "*The interests and welfare of the human being shall prevail over the sole interest of society or science.*" This provision further reinforces the importance of maintaining respect for human dignity and ensuring that an individual's rights are afforded paramount consideration. Accordingly, this might further be interpreted as applying to third party use of genetic testing and genetic information for economic objectives, and ensuring the primacy of individuals' fundamental rights in these circumstances. It is therefore clear that the overall rationale of the Convention is respect for the human being and acknowledgment of fundamental rights in this era of advancing science and technology. In addition to the emphasis on human dignity and integrity, the Convention also takes an evident non-discrimination approach to this area. The next section will further explore the application of the Convention to the area of advancing genetic science.

5.2 The Convention and genetic discrimination

Although the Convention is shaped in terms of bioethics generally, it does acknowledge the reality of advancing genetic science and the issues this gives rise to. In shaping this discussion, there are a number of articles of the Convention which merit particular consideration.

As regards privacy, the Biomedicine Convention has however been criticised as offering "*scanty standards on privacy-related issues.*"⁵⁴ Nevertheless, Article 10 on private life and the right to information is relevant. This article encompasses both the right to know, as well as the right not to know. Article 10(1) states that "*everyone has the right to respect for private life in relation to information about his or her health.*" Article 10(1) reflects the concept contained in Article 8 of the ECHR concerning the right to privacy.⁵⁵ This provision provides for the right to privacy in respect of one's health, which by implication encompasses one's genetic information. Therefore, although genetic information is not expressly referred to here, it can be implied that genetic information is included, by virtue of

⁵² Biomedicine Convention, Preamble

⁵³ Article 1. See also Elmar Doppelfeld, 'Council of Europe in the field of bioethics: the Convention on Human Rights and Biomedicine and other legal instruments' (2008) 34 *Intensive Care Medicine* 939

⁵⁴ Henriette Roscam Abbing 'The Convention on Human Rights and Biomedicine – An Appraisal of the Council of Europe Convention' (1998) 5 *European Journal of Health Law* 377, 383

⁵⁵ Explanatory Report to the Convention on Human Rights and Biomedicine, para 62

the fact that it constitutes a very personal and sensitive form of health information (and as was established in *S v Marper and the United Kingdom*⁵⁶).

Article 10(2) guarantees the right to know and the exception to this right, which is the right not to know. It states: “*Everyone is entitled to know information about his or her health. However, the wishes of individuals not to be so informed shall be observed.*” As has been highlighted in previous chapters, the right to know is a recognition of the individual as a person and “*an autonomous being.*”⁵⁷ Similarly, the right not to know is also an expression of one’s autonomy, and one’s potential desire not to receive information relating to health and particularly future health. The right not to know is particularly relevant in relation to genetic information, in light of its predictive value and the details it can potentially reveal about an individual and an individual’s family members.

As mentioned, the Biomedicine Convention acknowledges the developments in genetic science in recent years, and specifically refers to genetic testing.⁵⁸ Chapter IV addresses the human genome and sets out governing principles in this area. In highlighting the non-discrimination framework, Article 11 prohibits “*any form of discrimination against a person on grounds of genetic heritage.*”⁵⁹ This provision recognises the potential for genetic science to become a means of selection and discrimination.⁶⁰ The specific reference to ‘genetic heritage’ as grounds for discrimination is important in recognising not only the potential for abuse of genetic test results, but also the potential for abuse of genetic information gleaned from family history of disease.

However, the exact interpretation and impact of this clause is not particularly clear, and in this regard, the Convention has been criticised in failing to elaborate on the meaning of ‘genetic heritage.’⁶¹ In ascertaining the meaning and intention behind Article 11, Hendriks argues that Article 11 must be interpreted in light of the overall goals of the Biomedicine Convention, and that the intention of Article 11 was to expand the scope of Article 14 of the ECHR, and to provide protection against discrimination on the grounds of genetic characteristics, (which is not explicitly provided for in Article 14).⁶² In addition, Article 11 fails to provide comprehensive and adequate guidance or protection against the discriminatory use of genetic information by particular third parties, or under what circumstances this might be deemed to be unlawful. Therefore, although the

⁵⁶ *S. and Marper v. the United Kingdom* (No. 30562/04) [2008] ECHR 1581 4 December 2008)

⁵⁷ Roberto Andorno, ‘The Oviedo Convention: A European Legal Framework at the Intersection of Human Rights and Health’ (2005) 2 *Journal of International Biotechnology Law* 133, 139

⁵⁸ Explanatory Report, Convention to the Convention on Human Rights and Biomedicine, para 71- 73

⁵⁹ Biomedicine Convention, Article 11

⁶⁰ Explanatory Report, Convention to the Convention on Human Rights and Biomedicine, para 74

⁶¹ Aart Hendriks, ‘Protection against Genetic Discrimination and the Biomedicine Convention’ (2005) in Jan K.M. Gevers, Ewoud H. Hondius and Joep H. Hubben (eds) *Health Law, Human Rights and the Biomedicine Convention* (Koninklijke Brill N.V. 2005) 215

⁶² *Ibid* at 215

overall message is clear, there is a certain ambiguity in the interpretation and application of this provision.

Article 12 of the Convention addresses the use of genetic testing. It states that *“tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counselling.”*⁶³ This provision can be interpreted as meaning that interested third parties are not permitted to request genetic test results, whenever it does not have a health objective. Article 12 therefore implies that an insurer is not entitled to ask for a predictive genetic test to be carried out as a precondition for concluding an insurance contract or determining the terms of the insurance policy.⁶⁴

Likewise, it is prohibited for an employer to carry out predictive genetic testing as part of pre-employment medical examinations, unless it serves a health purpose for the individual. This provision recognises the limited right to know of employers. This means that in particular circumstances, when the working environment is hazardous and may have certain undesirable consequences for the health of the individual, predictive genetic tests may be carried out with the objective of improving the working environment. Accordingly, such tests should only be utilised in the interest of the individual's health.⁶⁵ Where use of genetic testing in employment and insurance does not have a health objective, it may have a *“disproportionate interference”* with the right to privacy, and contrary to the Convention.⁶⁶ The emphasis on the right to privacy is key in recognising an individual's fundamental rights, as carrying more weight than third parties' potential interest in genetic information and use of genetic testing. Article 12 also requires genetic counselling in case of predictive testing, as an additional safeguard.

Articles 10, 11 and 12 offer reasonably strong and comprehensive protections against misuse of genetic information and abuse of genetic technologies. Against a strong human rights framework, it recognises the need to protect one's privacy, as well as to prohibit any form of discrimination on the grounds of one's genetic heritage. In addition, Article 21 is relevant as it implies a rejection of ownership in one's genetic material, and states *“The human body and its parts shall not... give rise to financial gain.”*⁶⁷ This is in line with the submissions made in chapter 5, as regards property rights over genetic information and material. Before further

⁶³ Biomedicine Convention, Article 12

⁶⁴ Explanatory Report, Convention to the Convention on Human Rights and Biomedicine, paragraph 86

⁶⁵ Ibid, para 85

⁶⁶ Ibid, para 86. For further discussion, Aalt- Willem Heringa, 'International and National Legal Instruments' in Gerards, J.H., Heringa, A.W., Janssen, H.L. (eds) *Genetic Discrimination and Genetic Privacy in a Comparative Perspective* (Intersentia 2005) 39

⁶⁷ Biomedicine Convention, Article 21

evaluation of the Convention, the following section will give a brief overview of the Additional Protocol to the Convention on Human Rights and Biomedicine concerning genetic testing for health purposes.

5.3 Additional Protocol to the Convention on Human Rights and Biomedicine concerning genetic testing for health purposes

An Additional Protocol to the Convention on Human Rights and Biomedicine (“Additional Protocol”) concerning genetic testing for health purposes, was introduced in 2008.⁶⁸ It complements Chapter IV of the Biomedicine Convention in specifically addressing the human genome and genetic testing, and offers further guidance in relation to the use of such technologies.

The objective of the Additional Protocol is to provide a legal framework with the aim of upholding fundamental human rights in response to developments in genetic science and technology. The intention of this framework is to prevent the abuse of these technologies. It further highlights the rights based approach taken by the CoE in this area.⁶⁹ Similar to the rationale of the Biomedicine Convention, this Additional Protocol places an emphasis on the dignity and integrity of the person, as a prime consideration in response to advances in genetic science. In highlighting the objective of this instrument, the Preamble of the Additional Protocol refers to the importance and necessity for biomedical advances in improving quality of life and producing other benefits for individuals, thereby recognising the importance of advancing genetic science in society, and the impact this can have on public health and society in general.⁷⁰ The Preamble also refers to the importance of maintaining human dignity and other human rights in the course of such biomedical research. Indeed a number of other articles refer to the importance of the human being and one’s dignity and integrity as a person.⁷¹

Against the ethos of protecting human dignity and the importance of individuals’ fundamental rights, the Additional Protocol sets out principles with regard to the quality of genetic services, prior information and consent, as well as genetic counselling. An entire section is dedicated to consent,⁷² including the issue of genetic testing on persons not able to consent,⁷³ and the issue of genetic testing for the benefit of family members is specifically addressed in a number of

⁶⁸ Additional Protocol to the Convention on Human Rights and Biomedicine concerning genetic testing for health purposes (2008) (“Additional Protocol”)

⁶⁹ Laurence Lwoff, ‘Council of Europe adopts protocol on genetic testing for health purposes’ (2009) 17 *European Journal of Human Genetics* 1374

⁷⁰ *Ibid.* Additional Protocol to the Convention on Human Rights and Biomedicine concerning genetic testing for health purposes, Preamble.

⁷¹ Additional Protocol, Article 1 and Article 3

⁷² *Ibid.*, Article 9

⁷³ *Ibid.*, Articles 10, 11 12

provisions.⁷⁴ Therefore there is an evident emphasis on privacy rights as a means to protect individuals' rights. Article 2 covers genetic tests, "*which are carried out for health purposes ...*"⁷⁵ Key to this definition is that tests are performed for health purposes. Therefore, tests carried out in third party contexts, for example, by employers, for economic objectives, or other non-medical reasons are presumably not covered. Article 5 sets out the requirement that "*genetic services are of appropriate quality.*" This article also sets out further necessary criteria and incorporates the need for clinical and scientific validity.⁷⁶ The Additional Protocol also applies specifically to direct – to – consumer (DTC) genetic tests, and provides that a genetic test for health purposes may only be performed under "*medical supervision.*"⁷⁷

The Additional Protocol provides for privacy rights. Article 16 provides for the protection of private life and the right to information obtained by means of genetic testing. This article echoes the privacy provisions of the Biomedicine Convention and the ECHR. This Article encompasses the right to know, as well as the right not to know. Article 19 addresses the issue of genetic screening for health purposes and the circumstances under which it is deemed appropriate to be used.⁷⁸

It is clear from the above analysis that this Additional Protocol contains a comprehensive range of provisions designed to provide a rights based framework to address genetic advances. The Protocol has been framed to recognise the importance of allowing genetic science and technological innovation to flourish, for the benefit of all in society, while at the same time recognising the need to provide appropriate safeguards to uphold an individual's fundamental human rights. The following section will further evaluate this Additional Protocol and the Biomedicine Convention.

5.4 Evaluation

The Biomedicine Convention and the Additional Protocol are key instruments in this area and represent a strong moral framework, with fundamental human rights at the forefront. In adopting this approach, these instruments speak undoubtedly against any kind of discrimination based on genetic testing, and highlight the importance of maintaining privacy and dignity. They represent an official standard at the CoE level and serve as a guideline for Member States. These instruments also highlight the progression of human rights in the CoE, and the recognition of the need to respond to advances in science and technology.

⁷⁴ Ibid, Articles 13, 14, 15, 18

⁷⁵ Ibid, Article 2

⁷⁶ Ibid, Article 5

⁷⁷ Ibid, Article 7

⁷⁸ Laurence Lwoff, 'Council of Europe adopts protocol on genetic testing for health purposes' (2009) 17 European Journal of Human Genetics 1374

Although these instruments provide a valuable moral and human rights framework, one particular shortcoming can be identified. It also has limited value in light of the few ratifications, which reduces its overall impact. In the case of the Additional Protocol, as of January 2013, only six states have signed it, and only two of those have ratified, meaning that the Additional Protocol is not yet in force. Therefore, the practical results of the Convention and the Protocol are limited as few of the present EU Member States have ratified. In addition, in providing only for a reporting procedure, the Biomedicine Convention can be criticised in terms of legal effect and enforcement.⁷⁹

Nevertheless, in spite of this obvious weakness, it can be concluded that the expressive value of the Convention and its Protocol is admirable in addressing this area and providing a framework to discuss the core ethical and legal issues arising.

6. Recommendations of the Committee of Ministers

There are also a number of recommendations of the Committee of Ministers which merit mentioning in terms of further illustrating the CoE's moral framework in this area. This section aims to give a very brief overview of the relevant recommendations, with the objective of highlighting the CoE's commitment to addressing the challenging issues arising as science and technological innovation develop. Although these recommendations are non-binding, they are important in terms of offering guidance to Member States and setting a certain standard in a particular area, particularly from a moral and human rights standpoint.

There are four main recommendations that are particularly relevant. The first recommendation is on Prenatal Genetic Screening, Prenatal Genetic Diagnosis and Associated Genetic Counselling.⁸⁰ This recommendation focuses on the appropriate application of genetic services, with an emphasis on privacy, non-discrimination, as well as the need for genetic counselling. The second recommendation is a recommendation on Genetic Testing and Screening for Health Care Purposes.⁸¹ It provides guidelines in relation to the appropriate use of genetic services including equality of access and non-discrimination, data protection considerations, and respect for self-determination. These two recommendations reflect some of the core principles contained in the

⁷⁹ Henriette Roscam Abbing 'The Convention on Human Rights and Biomedicine – An Appraisal of the Council of Europe Convention' (1998) 5 European Journal of Health Law 377, 380

⁸⁰ Recommendation No. R (90) 13 of the Committee of Ministers to Member States on Prenatal Genetic Screening, Prenatal Genetic Diagnosis and Associated Genetic Counselling (Adopted by the Committee of Ministers on 21 June 1990 at the 442nd meeting of the Minister's Deputies)

⁸¹ Recommendation No. R (92) 3 of the Committee of Ministers to Member States on Genetic Testing and Screening for Health Care Purposes (Adopted by the Committee of Ministers on 10 February 1992 at the 470th meeting of the Minister's Deputies)

Biomedicine Convention and the ECHR, and they recognise the need to uphold fundamental human rights in this area.

In 1997 a recommendation on the Protection of Medical Data was introduced.⁸² It includes a number of provisions including the right to privacy in respect of medical data (including 'genetic data'). It is noted that this recommendation refers specifically to the category of 'genetic data.' It is particularly relevant in terms of the definition of genetic data, which is defined as any data of whatever type "*concerning the hereditary characteristics of an individual or concerning the pattern of inheritance of such characteristics within a related group of individuals,*" thereby encompassing family medical history. The fourth recommendation is on the impact of genetics on the organisation of health care services and training of health professionals.⁸³ The focus of this recommendation is on the correct use of genetic services, and the appropriate training and education of health care professionals in this area.

These recommendations are a further reflection of the CoE's moral imperative to address these challenging issues, from a human rights perspective. They provide important insights and policy guidelines that are necessarily intended to inform future regulatory endeavours. There is a clear focus on maintaining fundamental principles such as human dignity, equality and privacy, and an acknowledgement of the potential for discrimination.

7. Other relevant policy instruments

In 2012, the CoE Committee on Bioethics carried out a public consultation on a document on Predictivity, Genetic Testing and Insurance. With a focus on the insurance field, the objective of this consultation was to gather comments and suggestions from bodies representing a myriad of diverse areas concerned (including consumers, patients, insurance companies, geneticists and other medical professionals), on proposals and questions regarding the use of predictive health-related data (in particular genetic data). As highlighted in chapter 3, genetic information has the potential to influence insurance practices. In the absence of some form of appropriate regulatory response, there is a real potential for not only abuse of insurance practices for financial objectives, but also for breach of the right to privacy and the right not to be discriminated against. The CoE Committee on Bioethics is currently examining the submissions and the comments with a view to preparing a possible legal instrument. At the moment, it is unclear what form this instrument will take. It is expected that an

⁸² Recommendation No. R (97) 5 on the Protection of Medical Data (Adopted on 13 February 1997)

⁸³ Recommendation CM (Rec 2010) 11 of the Committee of Ministers to Member States on impact of genetics and training of health organisation of health care services and training of health professionals (Adopted by the Committee of Ministers on 29th September 2010 at the 1094th meeting of the Minister's Deputies)

instrument will be drawn up in late 2013/ early 2014 setting out guidelines for this area.⁸⁴

8. Conclusion

This chapter has provided an overview of the current human rights norms of the CoE and the regulation of genetic information. In applying the provisions of the ECHR to this emerging area, it shows how human rights can adapt and evolve with changes in society and science, and this is illustrated particularly in the relevant case law of the ECtHR in this area. This chapter has illustrated the growing awareness in the CoE of scientific and technological developments, and the subsequent challenges presented. It has acknowledged the need to respond to these advances and develop a regulatory response based on a core human rights framework.

This awareness is particularly reflected in the Biomedicine Convention (and the Additional Protocol), which addresses the ethical and legal dilemmas arising in this area and frames the issues from an ethical and human rights perspective. Although there are certain shortcomings and criticisms that can be observed with this Convention (and the Additional Protocol), it is an important step in the right direction, and provides valuable guidance in terms of tackling the contours of this debate. The various recommendations of the CoE are also reflective of its commitment to acknowledging human rights as science and technology advance. Most recently, the CoE Committee on Bioethics has acknowledged the need to address the particular area of insurance, and the impact of genetics and other predictive information on the industry. It has taken a proactive approach to this area in anticipating the dilemmas, instead of waiting for the issues to become widespread and problematic.

On examining the current European perspective in this area, it is submitted that the CoE's position is significant, in terms of highlighting the core human rights framework and providing a moral basis from which to consider the issues arising, as science continues to advance. At this point, from a human rights perspective, it is also necessary to refer to the growing connection between the CoE (ECHR in particular) and the EU, which will be further discussed in the next chapter.

This chapter acknowledges the forward- thinking approach taking by the CoE, as a response to advancing science and technology, which force society to evolve and re- frame core human rights. The following chapter will continue the chart and elucidate the current EU perspective. It will examine the EU human rights position, from the perspective of the EU Charter of Fundamental Rights, as well as the current reach of EU non- discrimination law and EU data protection law. It will evaluate how these regulatory frameworks might apply to genetic information and what protection they might provide, as well as identify potential gaps in this protection.

⁸⁴ http://www.coe.int/t/dg3/healthbioethic/default_en.asp (accessed 18 April 2013)

Chapter 9: The European Union and genetic information

1. Introduction

Against the regulatory background provided in part two of this thesis, this chapter will provide a comprehensive overview of the current regulatory position in the EU, and how it potentially applies to the regulation of genetic information. This chapter will evaluate the current position in the EU and assess whether this is adequate to protect against misuse of genetic information. In illustrating the EU human rights framework, this chapter will begin with an overview of the Charter of Fundamental Rights of the European Union (CFREU), its relevance in shaping EU human rights (and its relevance as an impetus to EU legislative action). The CFREU forms a core element of the EU human rights framework, and brings EU law further in line with the ECHR, as highlighted in chapter 8.

This chapter will include an analysis of the current EU position in relation to data protection law, including an overview of the current reform of data protection laws that is taking place. This chapter will also include an examination of the current EU non- discrimination position, with a focus on the relevant Directives which shape non- discrimination law. It will examine the reach of the non- discrimination framework in this area. This chapter will also highlight the relevant policy documents which shape the EU perspective on the regulation of genetic information.

2. Charter of Fundamental Rights of the European Union

It is noted that human rights are increasingly becoming a core part of EU law and policy. This is reflected in the legislative and policy endeavours of the EU, as well as the interpretation of human rights by the ECJ and the ECHR. In line with this trend towards integrating human rights into the EU constitutional framework, the first section of this chapter will examine the CFREU, and its relevance in shaping EU human rights law.¹ The CFREU is one of the most important sources of fundamental human rights in the EU. The CFREU was declared by the European Parliament, Council and Commission in December 2000.²

The CFREU merits discussion, not only in terms of forming a core element of the EU constitutional framework, but also its influence over the EU's activities. This

¹ For further discussion, see Allan Rosas, 'The European Union and Fundamental Rights Human Rights' in Catarina Krause and Martin Scheinin (eds) *International Protection of Human Rights: A Textbook* (2nd edn, Abo Akademi University Institute for Human Rights 2012) 481

² Charter of Fundamental Rights of the European Union of 18 December 2000 [2000] OJ C 364/01 ("CFREU")

section will explore the value of the Charter both before and after the Lisbon Treaty, (which has had a significant impact on this area). It will also look at the relevant provisions of the Charter and its application to this discussion.

2.1 Background

Prior to the introduction of the CFREU, there was an increasing acknowledgement of the necessity of facilitating the protection of human rights for all individuals. It has been observed that this is as a result of, amongst other things, “*the development of the doctrine of direct effect and the ever- expanding nature of EC competences beyond merely economic aims into other political and social fields.*”³ The original treaties, which focused on economic objectives, did not contain reference to human rights and their protection. The ECJ took on the role of recognising human rights and developing human rights law and, for the first time in 1969, the ECJ held that fundamental rights constitute a core element of the general principles and framework of the EU.⁴ Human rights were gradually becoming an important element of EU law and policy.

The introduction of the CFREU was representative of a distinct increase in human rights and their protection in the EU. The introduction of the CFREU was also effective in reformulating the goals of the EU to a certain degree and in increasing the focus on human rights. It emphasises the trend towards acknowledging social concerns and individual rights in addition to focusing on the economic harmony of the Union.⁵ The Charter has the potential to facilitate the progression towards “*a more rights- oriented system legally, as well as contribute in more symbolic terms to the development of an EU rights- culture.*”⁶ It has been described as “*a syncretic document,*” which provides rights from a variety of different sources, including the ECHR.⁷ Therefore the Charter creates more cohesion between EU human rights law and the ECHR framework. The following section will examine the legal status and effect of the Charter both before and after the Lisbon Treaty.

2.2 Legal Status of the Charter prior to the Lisbon Treaty

³ Christian Franklin, ‘The Legal Status of the EU Charter of Fundamental Rights after the Treaty of Lisbon’ (2010- 2011) 15 Tilburg Law Review 137, 139

⁴ Case C- 29/69 *Erich Stauder v City of Ulm* [1969] ECR- 419

⁵ Agustin Jose Menendez, ‘Chartering Europe: Legal Status and Policy Implications of the Charter of Fundamental Rights of the European Union’ (2002) 40 Journal of Common Market Studies 3 471, 472- 473

⁶ Francis G. Jacobs, ‘The EU Charter of Fundamental Rights’, in Anthony Arnall and Daniel Wincott (eds) *Accountability and Legitimacy in the European Union* (Oxford University Press 2003) 284- 285

⁷ Erica Szyszczak, ‘Antidiscrimination Law in the European Community’ (2008) 32 Fordham International Law Journal 2 Article 8 531, 540

It is noted that the Charter was drafted with the intention of becoming a binding legal document.⁸ However, much confusion surrounded the actual status of this instrument. There was no consensus as to this issue and therefore it was left open to be decided at a later stage.⁹ Prior to the Lisbon Treaty, the Charter was effectively a soft law instrument, with no binding influence. Despite its non-binding nature, it is observed that the Charter did play a certain role in EU law and policy development in a number of ways.¹⁰ For example, the ECJ and the Court of First Instance both referred to the provisions of the Charter in several cases.¹¹ The courts drew attention to the fact that although the Charter was not a legally binding instrument, “*its mere existence illustrated the importance of the rights it set out in the Community legal order.*”¹² The European courts therefore saw and used the Charter as a valuable source of human rights in the EU legal framework.¹³

From an EU political perspective, it is also pointed out that EU institutions saw the Charter as a valuable and practical instrument in the EU, offering direction in terms of the EU human rights framework.¹⁴ Similarly, the EU Annual Report on Human Rights (2002) referred to the necessity “*to draw practical lessons from the proclamation of the Charter, and to guide its conduct by the rights contained in it... any proposal for legislation will now be subject to an a priori compatibility check with the Charter.*”¹⁵ This reference to “*compatibility check*” is interesting, in highlighting the intended influence of the Charter on EU legislative endeavours. Accordingly, prior to the Lisbon Treaty, the Charter was an influential and significant form of soft law in the EU.

2.3 The Lisbon Treaty

The Charter was afforded an elevated status by the Lisbon Treaty, and it increased the value of this instrument and its influence in EU law. The Lisbon Treaty was signed on 13 December 2007 and entered into force on 1 January

⁸ Commission, ‘Communication on the legal nature of the Charter of Fundamental Rights of the European Union’ (COM) (2000) 644, at 5

⁹ See for example, Declaration no. 23 on the Future of the Union, para. 5, annexed to the Final Act adopting the Treaty of Nice [2001] OJ C80/01, 83. Christian Franklin, ‘The Legal Status of the EU Charter of Fundamental Rights after the Treaty of Lisbon’ (2010- 2011) 15 Tilburg Law Review 137, 141

¹⁰ Ibid at 141

¹¹ Case T-54/99 *Max.mobil Telekommunikation Service GmbH v Commission* [2002] ECR II-00313, paras 48 and 57 and Case C-540/03 *European Parliament v Council* [2006] ECR I-05769, para 38

¹² Case C-131/03 *P R.J. Reynolds Tobacco Holdings Inc. and Others v Commission* [2006] ECR I-07795, para 30

¹³ Christian Franklin, ‘The Legal Status of the EU Charter of Fundamental Rights after the Treaty of Lisbon’ (2010- 2011) 15 Tilburg Law Review 137, 141

¹⁴ Ibid. The Commission announced its intention to seek to ensure the compatibility of new EU legislative proposals with the provisions of the Charter. See Commission, ‘Memorandum on the Application of the Charter of Fundamental Rights of the European Union’ SEC (2001) 380/3 final.

¹⁵ European Union Annual Report on Human Rights, adopted by the Council on 21 October 2002

2009. It was a significant development in the field of human rights, in bringing about changes in the EU.¹⁶

From a human rights perspective, a significant provision of the Lisbon Treaty is Article 6(1), which changed the status of the CFREU.¹⁷ Under Article 6(1) Treaty on European Union (TEU),¹⁸ the Charter now has the same legal value as the Treaties, and the EU is now under an obligation to comply with the EU Charter “in all its activities.”¹⁹ EU Member States are also obliged to comply with the Charter when implementing law.²⁰ It has been asserted that this new provision “makes the Charter legally binding.”²¹ It has been further asserted that the “ratification of the Lisbon Treaty means that the EU Charter has now become a cardinal element of the Union’s body of “primary”, that is “constitutional” rules.”²² Further, in 2010 the Commission published a communication providing that the EU Charter must be taken into account when the EU is legislating.²³ Accordingly, there is strong support for the view that, in terms of the practical implications, it is clear that the CFREU will have a clear impact on the EU in the enactment of legislation.

The Lisbon Treaty therefore enhanced the value of the Charter, and added to the emphasis on human rights within the EU constitutional order. In this regard, it has been observed that the EU seems to have “undergone a true genetic transformation, evolving from a *sui generis* international organization, mainly focused on market integration, to an autonomous legal order protecting and promoting the rule of law within and outside its boundaries.”²⁴ In addition to influencing the legislative activities of the EU, the CFREU will also be an influential source of fundamental rights for the ECJ when interpreting and applying EU law, as has already been demonstrated. In the case of *Scheke &*

¹⁶ See generally, Wolfgang Weiss, ‘Human Rights in the EU: Rethinking the Role of the European Convention on Human Rights after Lisbon’ (2011) 7 European Constitutional Law Review 64

¹⁷ Sionaidh Douglas-Scott, ‘The European Union and Human Rights after the Treaty of Lisbon’ (2011) 11 Human Rights Law Review 4 645

¹⁸ Article 6 (1) Treaty on European Union states: “The Union recognizes the rights, freedoms and principles set out in the Charter of Fundamental Rights of the European Union of 7 December 2000, as adapted at Strasbourg, on 12 December 2007, which shall have the same legal value as the Treaties. The provisions of the Charter shall not extend in any way the competences of the Union as defined in the Treaties.”

¹⁹ Lisa Waddington, ‘The European Union and the United Nations Convention on the Rights of Persons with Disabilities: A Story of Exclusive and Shared Competences’ (2011) 18 Maastricht Journal 4 431, 433

²⁰ Ibid at 433

²¹ Xavier Groussot and Laurent Pech, ‘Fundamental Rights Protection in the European Union post Lisbon Treaty’ Policy Paper, Fondation Robert Schuman, European Issue No 173 (14 June 2010) at 2

²² Ibid at 2

²³ Communication from the Commission: Strategy for the effective implementation of the Charter of Fundamental Rights by the European Union COM (2010) 573 final

²⁴ Giacomo Di Federico, *The EU Charter of Fundamental Rights: From Declaration to Binding Instrument* (Springer 2010) Preface

Eifert, the CFREU was referred to as a binding instrument for the first time.²⁵ In the *Test- Achats* case, the CFREU and its influence was also referred to.²⁶

Although the Charter has been given additional status, it does not afford an independent basis for legislative action, but instead acts to inform the interpretation of the treaties and any EU activity. As reflected in Article 6(1), which provides that the Charter “*shall not extend in any way the competences of the Union*,” the Lisbon Treaty did not change or expand the existing competences of the EU.²⁷ It has been observed that the issue here should not focus on whether the CFREU bestows upon the EU additional legislative powers for the advancement of human rights, but rather it should focus on whether the existing powers and legislative competences of the EU “*will be re- oriented*” as a result of the EU Charter.²⁸ In other words, the CFREU may act to reflect the evolution of existing human rights, and to provide a framework within which to re-examine the interpretation of human rights in the EU. In this regard, it has also been noted that the Charter could be used to “*extend the existing rights and the protections against discrimination in new directions*.”²⁹ The next section will look at one element of this evolution of human rights, in terms of the protection of genetic information.

2.4 CFREU and genetic information

Having established the importance of the CFREU in the EU constitutional framework and its legal value, the following section will explore the application of the CFREU to the issue of genetic information, and discrimination issues in particular. In this regard, it is submitted that the CFREU has the potential to inform the interpretation of existing legislation, and it also has the potential to influence any potential EU level action in this area (although, as noted, it does not act as an independent legal basis for action).

As is the case with many human rights instruments in the area, it is noted that human dignity³⁰ and integrity³¹ are core principles in the CFREU. The concepts of human dignity and integrity are particularly important considerations in attempting to address advancing science and technology and the issues that

²⁵ Joined Cases C- 92/09 and C- 93/09, *Volker and Markus Schecke GbR & Hartmut Eifert v Land Hessen & Bundesanstalt für Landwirtschaft und Ernährung* 2010 ECR I-000, para 45- 46

²⁶ Case C- 236/09 *Association Belge des Consommateurs Test- Achats ASBL v Conseil des ministres* [2011] (1 March 2011) ECR I- 00773

²⁷ Article 6(1) TEU

²⁸ Bruno de Witte et al, ‘Legislating after Lisbon – New Opportunities for the European Parliament,’ European University Institute, Robert Schuman Centre for Advanced Studies (Florence, June 2010)

²⁹ Available at [www.eui.eu/Projects/EUDO/Documents/EUDO-Legislatingafterlisbon\(SD\).pdf](http://www.eui.eu/Projects/EUDO/Documents/EUDO-Legislatingafterlisbon(SD).pdf) (accessed 25 June 2013)

²⁹ Erica Howard, ‘EU Equality Law: Three Recent Developments’ (2011) 17 *European Law Journal* 6785, 788

³⁰ CFREU, Article 1

³¹ CFREU, Article 3

arise. These provisions are also reflective of some of the Council of Europe norms in this area, as well as some of the international soft law instruments, as previously discussed.

In line with international and Council of Europe norms, the Charter places a certain emphasis on privacy and data protection. Article 7 provides for respect for private and family life.³² Article 8 on the protection of personal data states: *“Everyone has the right to the protection of personal data concerning him or her.”*³³ The right to protection of personal data is a key consideration and a fundamental right in the EU (as discussed in chapter 5). In light of the sensitive and personal nature of genetic information and the details it reveals for the individual and family members, these are relevant provisions. It certainly confirms the importance and primacy of the right to privacy and the concept of data protection in the EU human rights framework. As highlighted in chapter 5, the data protection model is an expression of the right to privacy.

The concept of non- discrimination is also firmly reflected in the Charter. The Charter includes Article 21(1) which is a key provision for the purposes of this debate. Article 21(1) provides: *“Any discrimination based on any ground such as ... genetic features ... disability, ... shall be prohibited.”* This is a broadly drafted provision prohibiting discrimination on a range of grounds. The specific reference to ‘genetic features’ is a groundbreaking step in the protection of human rights in the EU and indicates both the evolution of human rights and a commitment by the EU to acknowledge the importance of protecting ‘genetic features.’ It is submitted that (unjustified) discrimination by employers, insurance companies, and other interested third parties on the grounds of ‘genetic features’ may therefore be contrary to Article 21(1). It is asserted that Article 21(1) might shape and inform the interpretation of other pieces of EU legislation, for example the Framework Employment Directive. It might also influence the activities of the EU if and when enacting legislation in this area, in line with the *“compatibility check,”* as noted above.

Another relevant article in this debate is Article 26 on the integration of persons with disabilities. It states: *“The Union recognises and respects the right of persons with disabilities to benefit from measures designed to ensure their independence, social and occupational integration and participation in the life of the community.”*³⁴ This provision recognises the rights and freedoms of persons with disabilities. Specifically, it acknowledges the importance of inclusion in society, and recognises the rights of persons with disabilities in this regard. In the context of this discussion, misuse of genetic information by third parties might operate to create barriers to employment and social goods such as insurance, which might have a particularly adverse effect on persons with disabilities (and persons with putative disabilities), and may further inhibit participation in society.

³² CFREU, Article 7

³³ CFREU, Article 8

³⁴ CFREU, Article 26

By way of highlighting the scope of the CFREU, and illustrating the reach of fundamental human rights, Article 35 on health care also merits mentioning. Article 35 states “*Everyone has the right of access to preventive health care and the right to benefit from medical treatment under the conditions established by national laws and practices. A high level of human health protection shall be ensured...*”³⁵ As highlighted, genetic technologies are becoming increasingly available to individuals, offering the possibility of individuals being better informed as to their genetic status and health. However, there are corresponding risks that may create barriers to accessing genetic services, such as fear of discrimination and other misuse of genetic information. In order to eliminate this fear, and instil confidence in individuals, appropriate regulation is necessary, allowing access to enhanced health care and scientific advances for all individuals. Public confidence in science and new technologies is needed in order for science to further progress. Article 35 reinforces this submission.

Based on the above analysis, the Charter therefore provides a clear human rights framework within which to view this area. It is also submitted that the Charter has the potential to shape EU level regulation of genetic information. In this regard, it is submitted that the EU must have regard to the Charter when enacting legislation. The following section will further explore the influence that the Charter might have on the regulation of genetic information.

2.5 Can the CFREU shape EU regulation of genetic information?

The additional value of the Charter (post Lisbon) and the specific reference to ‘genetic features’ can arguably strengthen arguments in support of the regulation of this area and perhaps prompt the EU to reflect upon the case for EU level regulation of genetic information. Although it is emphasised that it does not extend the competences of the EU, at the very least, the Charter can inform the interpretation of existing legislation to take account of advancing genetic science, and the corresponding need to recognise the protection of human rights. It is a core human rights instrument in the EU and highlights the commitment of the EU to recognising the fundamental human rights of all individuals.³⁶ The interpretation of human rights is evolving as society, science and technology advance, as is illustrated in international human rights law. Human rights in the EU legal framework are similarly evolving and it is submitted that the CFREU has the potential to drive this evolution and expand the interpretation of human rights, in response to advancing genetic science.

In the context of highlighting the increasing cohesion between the EU and the ECHR, Article 6(2) is relevant in providing that the EU “*shall accede*” to the

³⁵ CFREU, Article 35

³⁶ Maria Eduarda Gonclaves and Maria Ines Gameiro, ‘Does the Centrality of Values in the Lisbon Treaty Promise More Than it Can Actually Offer: EU Biometrics Policy as a case study’ (2013) (first published online 4 June 2013) 1, 12

ECHR. This inevitable future accession of the EU to the ECHR will further highlight the importance of the ECHR in the EU legal order.

Having established the core human rights framework in the EU, and the application to this area, the following sections will examine the EU data protection position and the non-discrimination position, and explore the reach of these frameworks to the regulation of genetic information.

3. EU Data Protection legislation

As discussed in chapter 5, the concept of data protection is an expression of an individual's right to privacy. Specifically, data protection law protects the privacy of one's personal information by maintaining control over the disclosure and use of such information. The protection of an individual's personal data is a well-established principle in EU law, pursuant to Article 16 TFEU.³⁷ Article 16 establishes a clear basis to regulate the processing of personal data in the EU. In addition, the right to privacy and the right to protection of personal data are provided for in Articles 7 and 8 of the CFREU, as highlighted above.³⁸ The provisions in the CFREU strengthen the emphasis on the protection of personal data in the EU and enhance the overall human rights framework in this area. This section will discuss the Data Protection Directive and will highlight the recent developments in the area which point to an intention to reform data protection laws in the EU.

3.1 Data Protection Directive

In 1995 the EU introduced a directive addressing data protection with the intention of ensuring the right of individuals to control the uses to which their personal data is put.³⁹ Within a clear privacy framework, the purpose of the Data Protection Directive is to harmonise data protection law, facilitating the development of the EU internal market while at the same time enhancing public confidence. This directive was designed to protect the privacy and protect all personal data collected for or about citizens of the EU.⁴⁰ It ensures the flow of personal information, while at the same time ensuring that individuals maintain

³⁷ TFEU, Article 16. See generally Marc Rotenberg and David Jacobs, 'Updating the law of information privacy: the new framework of the European Union' (2013) 36 Harvard Journal of Law and Policy 2 605

³⁸ CFREU, Article 7 and Article 8

³⁹ Council and Parliament Directive 95/46/EC of 24 October 1995 on the protection of individuals with regard to the processing of personal data and on the free movement of such data [1995] OJ L 281/31 ("Data Protection Directive")

⁴⁰ Marc Rotenberg and David Jacobs, 'Updating the law of information privacy: the new framework of the European Union' (2013) 36 Harvard Journal of Law and Policy 2 605, 617

control over the use of personal data.⁴¹ In confirming the harmony between the EU and the CoE in this area, the Directive notes that the rights contained therein are reflective of those in the Convention for the Protection of individuals with regard to Automatic Processing of Personal Data.⁴²

Data protection law is an effective means of securing the right to privacy and other fundamental human rights, such as the right to autonomy. The Data Protection Directive is therefore recognised as a significant element of EU human rights law and privacy law.⁴³ It is in line with the privacy and data protection provisions in the CFREU, and the overall EU human rights framework. The following sections will examine the specific provisions contained in the Data Protection Directive.

A key concept contained in the Directive is that of 'personal data.' Personal data is defined as '*any information relating to an identified or identifiable natural person ('data subject')*'.⁴⁴ An identifiable person is "*one who can be identified, directly or indirectly, in particular by reference to an identification number or to one or more factors specific to his physical, physiological, mental, economic, cultural or social identity*."⁴⁵ In the context of this discussion, the question arises whether genetic information constitutes 'personal data' for the purposes of the Directive. Although potentially genetic information could be interpreted as 'personal data', this has not been interpreted or confirmed as such.⁴⁶

This Directive provides a number of strong protections with the objective of safeguarding an individual's data and facilitating control over the disclosure and use of data. The Directive dictates that personal data must be collected with proportionality, and necessitates the need for the consent of the person to whom the data is related.⁴⁷ For example, it provides that a data controller⁴⁸ ensure that personal data is, *inter alia*, processed fairly and lawfully.⁴⁹ There are relatively strong protections in respect of an individual's personal data, and the legislation places strict obligations on third parties. Article 7 stipulates the criteria necessary

⁴¹ Andrew Charlesworth, 'Information Privacy in the European Union: E Pluribus Unum or Ex Uno Plures? (2002- 2003) 54 Hastings Law Journal 931, 931. See also Maeve Mc Donagh, 'Balancing Disclosure of Information and the Right to Respect of Private Life in Europe' (2012) Journal of Internet Law 3, 5

⁴² Convention for the Protection of individuals with regard to Automatic Processing of Personal Data (1981, ETS 108)

⁴³ Olivier De Shutter, 'Three Models of Equality and European Anti- Discrimination Law' (2006) 57 Northern Ireland Legal Quarterly 1 25

⁴⁴ Data Protection Directive, Article 2 (a)

⁴⁵ Ibid

⁴⁶ Taylor has noted the uncertainty surrounding the concept of personal data and its meaning. See Mark Taylor, *Genetic Data and the Law: A Critical Perspective on Privacy Protection* (Cambridge University Press 2012) 78

⁴⁷ Data Protection Directive, Article 6

⁴⁸ Ibid, Article 2 (d)

⁴⁹ Ibid, Article 6

in order to ensure that the processing of data is legitimate.⁵⁰ For example, it is necessary that “*the data subject has unambiguously given his consent.*”⁵¹ This ensures that the individual maintains a level of control over the personal data, and it safeguards the right to privacy. There are a number of other criteria set out, for example, where processing of personal data is necessary to protect the vital interests of the data subject⁵² or where such processing is necessary in the public interest.⁵³

The Directive has a wide scope of application and applies, *inter alia*, in the employment and insurance contexts. For example, in the context of data protection in the insurance context, the EU data protection legislation is applicable, in terms of protecting the insurance customer’s personal information. Likewise, in the employment context, an individual’s personal information is subject to the provisions set out above, to ensure control over such information. Therefore as regards the protection of the privacy of personal information in the employment and insurance fields, there is a reasonably effective regulatory regime in place. The question then arises whether genetic information is encompassed within the concept of personal data. In this regard, it is necessary to examine the sensitive categories of data that are provided for in the Directive.

3.1.1 Special categories of data

The Directive provides that if personal information reveals ‘racial or ethnic origin, political opinions, religious or philosophical beliefs, trade- union membership’ or concerns ‘health,’ then it will fall within the category of personal data that is deemed sensitive and subject to additional conditions of ‘fair processing.’⁵⁴ The Directive establishes a presumption against the processing of any sensitive personal data. The presumption can, however, be rebutted according to certain specified circumstances,⁵⁵ for example, if the data subject has given ‘explicit consent’ to the processing of the sensitive personal data.⁵⁶

In the context of this debate, as noted in chapter 3, genetic information may potentially reveal sensitive details about an individual’s ‘racial or ethnic origin’, and indeed ‘health or sex life,’ and accordingly may be deemed to be sensitive personal data and subject to additional conditions of ‘fair processing.’ This issue has not however been interpreted by the European courts, so it is not certain how such a question might be interpreted. In addition, although the Directive does not explicitly refer to genetic data, it does mention health data, which is deemed to

⁵⁰ Ibid, Article 7

⁵¹ Ibid, Article 7 (a)

⁵² Ibid, Article 7 (e)

⁵³ Ibid, Article 7 (f)

⁵⁴ Ibid, Article 8

⁵⁵ Mark Taylor, *Genetic Data and the Law: A Critical Perspective on Privacy Protection* (Cambridge University Press 2012) 83

⁵⁶ Data Protection Directive, Article 8 (2)(a)

be a special category of data.⁵⁷ There is currently no substantive provision for genetic data and no specific reference to this category of information. Under the 1995 Directive, there is therefore a clear gap in the protection for genetic data. This provides the introduction for the next section which examines the recent developments in this area, to address the evident gaps in protection.

3.2 Recent developments in EU Data Protection law

Although the 1995 Directive was a milestone in the history of data protection and an important recognition at EU level of data protection as a fundamental human right, there was a general consensus that the legislation needed to be updated, in line with modern society and advancing technology.⁵⁸ Indeed, recent developments in this area point to an intention to include genetic data within the ambit of EU data protection laws. In 2012, the Commission proposed a major reform of the EU legal framework on the protection of personal data. The new proposals will strengthen individual data protection rights and address the challenges of new technologies and changing society. It is a recognition of the reality of modern society and advancing technology, as well as a recognition of the potential for misuse of personal information if not sufficiently protected. It recognised the inadequacies and gaps in the current legislation and the need to reform the law.⁵⁹

This section will trace the history behind this data protection reform process. The intention to modernise data protection laws was initiated several years ago. In March 2004 the EU's Article 29 Data Protection Working Party claimed that genetic data has unique characteristics compared to health data and therefore merits additional legal protection.⁶⁰ The document refers to Article 21(1) of the CFREU, noting the prohibition of discrimination based on genetic features. The document also noted that genetic data should not be seen in a reductionist way, with reference to the concept of genetic determinism. The overall tenor of the document is one of genetic exceptionalism, and it recognises the unique nature of genetic information, as well as the potential for misuse. Further developments came in 2004 when the European Commission produced a document with 25 Recommendations on the Ethical, Legal and Social Implications of Genetic Testing (which will be discussed in the final part of this chapter).⁶¹ These

⁵⁷ Ibid, Article 8 (1)

⁵⁸ European Union Article 29 Data Protection Working Party Working Document on Genetic Data (March 2004) (12178/03/EN WP 91)

⁵⁹ Marc Rotenberg and David Jacobs, 'Updating the law of information privacy: the new framework of the European Union' (2013) 36 Harvard Journal of Law and Policy 2 605, 623

⁶⁰ European Union Article 29 Data Protection Working Party Working Document on Genetic Data (March 2004) (12178/03/EN WP 91)

⁶¹ European Commission 25 Recommendations on the Ethical, Legal and Social Implications of Genetic Testing, (European Commission Expert Group, 2004). Available at: http://ec.europa.eu/research/conferences/2004/genetic/pdf/recommendations_en.pdf (assessed 31 January 2013)

developments paved the way in highlighting the area of advancing genetic science as one that merits protection.

In December 2010 the European Commission introduced a Communication calling for, *inter alia*, genetic data to be expressly defined and provided for. It was stated that “*In the light of technological and other societal developments, there is a need to reconsider the existing provisions on sensitive data, to examine whether other categories of data should be added and to further clarify the conditions for their processing. This concerns, for example, genetic data which is currently not explicitly mentioned as a sensitive category of data.*”⁶² Then in July 2011 the European Parliament adopted a Resolution on a comprehensive approach on personal data protection in the EU.⁶³ It was stated that the European Parliament “*supports further clarification ... on the processing of sensitive data, and calls for reflection on the need to deal with new categories such as genetic and biometric data ...*”⁶⁴

On 25 January 2012, the European Commission introduced a draft European Data Protection Regulation – ‘*Regulation of the European Parliament and of the Council on the protection of individuals with regard to the processing of personal data and on the free movement of such data.*’⁶⁵ With these new Regulations, the Commission aims to develop an updated and unified data protection framework throughout the EU. It is also evident that the Commission is committed to reform and modernise data protection legislation, in line with the realities of today’s society and changing norms.⁶⁶ The following sections will highlight some of the main changes proposed.

3.2.1 Major changes of the regulations

Importantly, the draft Regulations identify ‘genetic data’ as a category of personal data designated for special protection. ‘Genetic data’ is defined broadly to include “*all data, of whatever type, concerning the characteristics of an individual that are inherited or acquired during early prenatal development,*” therefore presumably incorporating all genetic information as well as family medical history and other health related information.⁶⁷ This broad definition and the implication of

⁶² Communication from the Commission: A Comprehensive approach on personal data protection COM (2010) 609 final

⁶³ European Parliament Resolution of 6 July 2011 on a comprehensive approach to personal data in the European Union (2011/2025)

⁶⁴ Ibid

⁶⁵ Regulation of the European Parliament and of the Council on the protection of individuals with regard to the processing of personal data and on the free movement of such data (General Data Protection Regulation) COM (2012) 11 final

⁶⁶ Marc Rotenberg and David Jacobs, ‘Updating the law of information privacy: the new framework of the European Union’ (2013) 36 Harvard Journal of Law and Policy 2 605, 623

⁶⁷ Regulation of the European Parliament and of the Council on the protection of individuals with regard to the processing of personal data and on the free movement of such data (General Data Protection Regulation) COM (2012) 11 final, Article 4 (10)

encompassing family medical history is certainly welcomed, and it will undoubtedly operate to increase the protection afforded on the grounds of genetic data, and also close some existing gaps in national level legislation.⁶⁸ In addition, the proposed legislation will also operate to strengthen the EU's internal market, by creating more cohesion and harmony between Member States.

As regards protection of genetic privacy in the context of employment, insurance (and other third party contexts), these draft Regulations are to be welcomed as explicitly recognising genetic data as a category that deserves protection and merits privacy safeguards. It is also recognition of the realities of advancing genetic science. Robust data protection laws in this area will operate to protect sensitive genetic information, thereby protecting an individual's (and family members') autonomy, as well as preventing unwanted disclosure to third parties. Accordingly, there will be certain obligations on employers, insurers and other third parties as regards handling and using genetic data. The draft Regulations will have the effect of strengthening the EU's data protection and privacy standards. The following section will address the efficacy of this new regime in this area.

3.3 Is data protection legislation sufficient to fully protect genetic information?

The question then arises whether strong EU data protection legislation is sufficient to fully protect EU citizens against misuse of genetic information. Strong data protection laws would safeguard an individual's privacy rights. As highlighted in chapter 5, by maintaining control over the disclosure and use of personal information, such laws also have the effect of safeguarding other fundamental rights, such as the right to dignity and autonomy. From a human rights perspective, there are compelling reasons in favour of such data protection laws as a means of protecting genetic information. The proposed legislation would seem to provide adequate protection for the disclosure and processing of genetic data.

However, it is questionable whether this framework would sufficiently and fully protect against other misuse, such as the discriminatory use of genetic information. To a certain extent, data protection laws only provide limited protections in these circumstances, as is highlighted in chapter 5. For example, such laws would not offer protection against the discriminatory use of genetic information. Therefore, once the individual has consented to the disclosure of genetic information, or if it is in the public domain, there is still a potential that such information will be misused. This may point to the need for additional protections.

⁶⁸ For example, in Ireland, there are gaps in the protection available under Part 4 of the Disability Act 2005, as regards use of family medical history.

In light of advancing genetic science and the increasing availability of genetic information, and in light of the above-mentioned shortcomings with the data protection framework, it is unlikely that this one-dimensional approach would fully protect EU citizens against the misuse of genetic information in the long term. For now though, it is certainly a welcome endeavour, and one which will go some way towards protecting sensitive genetic information and instilling confidence in individuals. These questions will be further explored and developed in chapters 10 and 11.

4. EU non-discrimination legislation

This section will explore the EU non-discrimination framework and will highlight the rights based approach taken to this area. The field of non-discrimination law in the EU is comprised primarily by EU Directives, which promote integration and uniformity. The EU non-discrimination framework has only been developed in recent years, with the first legislative endeavours in 2000.⁶⁹ Accordingly, this section will focus primarily on the relevant directives that have been adopted in this area. This section will also illustrate how non-discrimination law has been interpreted by the ECJ, particularly on the ground of disability and how case law has helped to shape the current legal position. It will examine the reach of current non-discrimination law and specifically the extent to which it might apply to discrimination on the grounds of genetic predisposition to disability. In addition to the current EU non-discrimination law, this section will also highlight the proposed new non-discrimination Directive, which, if ever introduced, has the potential to further shape the EU non-discrimination framework.

4.1 The 'Article 13 Directives'

Article 13 (now Article 19) in the EC Treaty at Amsterdam granted the EU the authority to prohibit discrimination on a number of grounds, including race and ethnic origin, sexual orientation, religion and belief, age and disability. This was a new departure for the EU and presented new opportunities in the field of non-discrimination law and the protection of human rights. In 2000 Article 13 provided the legal basis for a Directive tackling discrimination in the area of employment on a range of grounds, including disability.⁷⁰ It also acted as a legal basis for the Race Directive, introduced in 2000 and the Gender Equal Treatment Directive, introduced in 2004.⁷¹ These three 'Article 13 Directives' will be discussed in this

⁶⁹ Grainne de Burca, 'The Trajectories of European and American Antidiscrimination Law' (2012) 60 *American Journal of Comparative Law* 1, 8

⁷⁰ Council Directive 2000/78/EC of 27 November 2000 establishing a general framework for equal treatment in employment and occupation [2000] OJ L 303/16 ("Framework Directive"). See Lisa Waddington, 'A New Era in Human Rights Protection in the European Community: The Implications the United Nations Convention on the Rights of Persons with Disabilities for the European Community' (2007) 4 *Maastricht Faculty of Law Working Paper*, 1, 9

⁷¹ Council Directive 2000/43/EC of 29 June 2000 implementing the principle of equal treatment between persons irrespective of racial or ethnic origin [2000] OJ L 180/22 ("the Race Directive"),

section, highlighting the commitment by the EU to addressing issues of non-discrimination, on a variety of grounds. In highlighting the commitment to upholding fundamental human rights, it is observed that the 'Article 13 Directives' "*adopt the classic, human rights model to combating discrimination: individual and rights based.*"⁷²

4.2 The Employment Equality Directive

Of primary relevance in the context of non-discrimination in the area of employment is the Employment Equality Directive (the Framework Directive).⁷³ The Framework Directive was the Community's "*first rights- conferring instrument for persons with disabilities*"⁷⁴ and the Commission referred to it as a "*path-breaking piece of legislation.*"⁷⁵ The Directive promotes the concept of equal treatment, and prohibits discrimination on a number of grounds, including disability.⁷⁶ It covers the fields of employment and occupation, vocational training, membership of employer and employee organisations.

In terms of the framework used, it places the disability ground (as well as the other grounds of discrimination) in a strong human rights based framework, thereby moving away from the traditional view of seeing persons with disabilities as the recipients of benefits towards the modern view of persons with disabilities as rights holders entitled to equal opportunities on an equal basis with others. The Framework Directive uses the tool of non-discrimination to achieve this objective and uphold a person's human rights. The ECJ case of *Mangold* reaffirms the human rights objective underlying the Framework Directive.⁷⁷

In line with human rights discourse, Quinn links the merits of the Framework Directive to the CFREU, and highlights the two main provisions in the Charter which mention disability.⁷⁸ As noted, Article 21(1) of the CFREU prohibits

Council Directive 2004/113/EC of 13 December 2004 implementing the principle of equal treatment between men and women in the access to and supply of goods and services [2004] OJ L 373/37 ("the Gender Equal Treatment Directive")

⁷² Catherine Barnard, *European Community Employment Law* (3rd edn, Oxford University Press 2006) 311

⁷³ The Framework Directive

⁷⁴ Richard Whittle, 'The Framework Directive for Equal Treatment in Employment and Occupation: An Analysis from a Disability Rights Perspective' (2002) 27 *European Law Review* 303, 305

⁷⁵ Commission Staff Working Paper: Disability Mainstreaming in the European Employment Strategy, at 3, COM (2005) EMCO/11/ (1 July 2005)

⁷⁶ See generally, Lisa Waddington, 'When is it Reasonable for Europeans to be Confused: Understanding when a Disability Accommodation is "Reasonable" from a Comparative Perspective' (2007- 2008) 29 *Comparative Labor Law and Policy Journal* 317. See also Gerard Quinn, 'Disability Discrimination Law in the EU', in Helen Meenan (ed) *Equality Law in an Enlarged European Union – Understanding the Article 13 Directives* (Cambridge University Press 2007) 242

⁷⁷ Case C- 144/04 *Mangold v Helm* [2005] ECR I- 9981

⁷⁸ Gerard Quinn, 'Disability Discrimination Law in the EU' in Helen Meenan (ed) *Equality Law in an Enlarged European Union – Understanding the Article 13 Directives* (Cambridge University Press 2007) 242

discrimination on a number of grounds including disability and genetic features, and Article 26 of the Charter focuses on the integration of persons with disabilities. The additional value added to the Charter further reinforces the strength of these provisions, and further complements the objective and impact of the Framework Directive. The Framework Directive is therefore situated within a firm human rights based paradigm, with individual rights at the fore. The following sections will explore the potential scope and application of the Framework Directive.

4.2.1 Definition of discrimination under the Framework Directive

In the context of examining the merits of the Framework Directive, and in evaluating this legislation within the context of potentially regulating genetic information, it is necessary to examine the definition of discrimination. Article 2 addresses the concept of discrimination. It states that “*the principle of equal treatment*” means “*that there shall be no direct or indirect discrimination whatsoever on any of the grounds referred to in Article 1.*”⁷⁹ Direct discrimination under the Directive is defined as “*where one person is treated less favourably than another is, has been or would be treated in a comparable situation, on any of the grounds ...*”⁸⁰

Indirect discrimination has been defined as “*where an apparently neutral provision, criterion or practice would put persons having ... a particular disability... at a particular disadvantage compared with other persons ...*”⁸¹ The concept of discrimination under the Framework Directive is therefore broadly drafted to cover situations of blatant less favourable treatment, as well as more covert disadvantageous treatment that may have a particularly adverse effect on a certain individual or group.

Indirect discrimination may sometimes be provoked by prejudice and other negative attitudes. In other words, historical prejudice and stigma directed towards persons with disabilities may result in some employers adopting indirect strategies or policies to eliminate such individuals from employment pools. As has been illustrated in chapter 5, the concept of ‘indirect discrimination’ might be construed to apply to genetic discrimination, in certain circumstances. For example, it is established that certain genetic conditions are more prevalent in certain ethnic minorities and amongst the sexes. An employer may therefore take advantage of existing inequities and target certain groups with strategic policies or practices, based upon negative attitudes and perceptions towards disability (and genetic predisposition to disability).

⁷⁹ Framework Directive, Article 2 (1)

⁸⁰ Ibid, Article 2(2)(a)

⁸¹ Ibid, Article 2(2)(b)

Having set out the scope of discrimination under the Framework Directive, the next question relates to the definition of disability, which will be discussed in the next section.

4.2.2 Definition of disability

It is acknowledged that there is no definition of disability provided for in the Framework Directive. Quinn has analysed the absence of a definition of disability. In this regard he refers to Article 1 and its impact, which states that the general purpose of the Directive “*is to lay down a general framework for combating discrimination on the ‘grounds of’ disability with a view to putting into effect the principle of equal treatment.*”⁸² Here, it is the discrimination or the negative treatment ‘on the ground’ of disability that is proscribed.⁸³ This concept is reflective of the social model of disability. It takes the focus away from the individual, and looks at the act of discrimination or negative treatment.

In the context of this debate, individuals who may be predisposed to future disability, such as those with a family history of illness or those who test positive for a certain gene, may be adversely treated or discriminated against ‘on the basis of disability.’⁸⁴ In practical terms, an individual who tests positive for having a predisposition to breast cancer, heart disease or any other genetically based disease, and who is therefore not disabled, may well be treated adversely by employers (and other third parties) as if they were disabled. Using the logic and rationale of discrimination ‘on the grounds of disability,’ then those with genetic susceptibilities may well come within the scope of non- discrimination law.⁸⁵ However, although in theory it may make sense to view discrimination ‘on the basis of disability’ in this broad manner, and although it is clear that, to a certain extent the Framework Directive is reflective of the social model of disability, the question has been left open to the courts to interpret. The following section will explore the case law of the ECJ and how it has shaped the interpretation of disability.

4.2.3 Chacon Navas v Eurest Colectividades SA

This section will give an overview of the relevant case law of the ECJ and its application. In particular, it will illustrate the evolution of the ECJ case law in this area, from the initial narrow approach to the recent, welcomed interpretation of disability in line with current thinking. It will analyse the cases

⁸² Ibid, Article 1

⁸³ Gerard Quinn, ‘Disability Discrimination Law in the EU’ in Helen Meenan (ed) *Equality Law in an Enlarged European Union – Understanding the Article 13 Directives* (Cambridge University Press 2007) 248

⁸⁴ Ibid

⁸⁵ Ibid at 250- 251

of *Chacon Navas*, and *Coleman v Attridge Law* as seminal cases on this issue. It will also highlight the recent cases of *Ring and Werge*, and *Commission v Italy*.

An instrumental case is the *Chacon Navas* case.⁸⁶ The *Chacon Navas* case has attracted criticism and is unfortunate particularly in light of the restrictive view taken on the definition of disability.⁸⁷ In the *Chacon Navas* case, the ECJ was called upon to decide if the employee, who was absent from work for some time as a result of illness, was to be regarded as a person with a disability for the purposes of the Directive.

The court found that current medical conditions that may indicate future disabilities do not bring the individual within the ambit of the Directive.⁸⁸ The court defined disability as a “*limitation which results in particular from physical, mental or psychological impairments and which hinders the participation of the person concerned in professional life*.”⁸⁹ Pursuant to this definition, the source of the ‘limitation’ is the ‘impairment’ inherent in the individual, and this ‘impairment’ causes or impacts upon an individual’s inclusion and participation in professional life, thereby reflecting the medical construction of disability. Accordingly, this definition does not see society or societal barriers as the problem, but instead views the problem as being within the individual.⁹⁰

This decision has attracted a great deal of criticism as endorsing the medical model of disability. This formulation is also out of line with the rationale and objectives of the CRPD, and the social model of disability.⁹¹ It has also been observed that the court’s rationale is arguably “*uninformed by the historical, political, and normative debate regarding the meaning of disability in Europe and abroad*.”⁹² In the context of this debate and in an age of advancing genetic science, the court’s decision in this case was disappointing, and created a gap in protection for individuals with genetic predispositions and putative disabilities.

⁸⁶ Case C-13/05 *Chacon Navas v Eurest Colectividades SA* [2006] ECR I-6467

⁸⁷ Lisa Waddington, ‘Case C-13/05, *Chacon Navas v Eurest Colectividades SA*, Judgment of the Grand Chamber of 11th July 2006, (2007) 44 Common Market Law Review, 487. For review of case, see Gerard Quinn, ‘Disability Discrimination Law in the EU’ in Helen Meenan (ed) *Equality Law in an Enlarged European Union* (Cambridge University Press 2007)

⁸⁸ *Ibid*

⁸⁹ Case C- 13/05 *Chacon Navas v Eurest Colectividades SA* [2006] E.C.R. I-6467, para 62

⁹⁰ Lisa Waddington, ‘*Breaking New Ground: The Implications of Ratification of the UN Convention on the Rights of Persons with Disabilities for the European Community*’ in Oddný Mjöll Arnardóttir and Gerard Quinn (eds) *UN Convention on the Rights of Persons with Disabilities* (Martinus Nihoff 2009) 133. For further discussion of the *Chacon Navas* case, see Gerard Quinn, ‘Disability Discrimination Law in the EU’ in Helen Meenan (ed) *Equality Law in an Enlarged European Union* (Cambridge University Press 2007) 257. See also Lisa Waddington, ‘Case C-13/05, *Chacon Navas v. Eurest Colectividades SA*, Judgment of the Grand Chamber of 11th July 2006’ (2007) 44 Common Market Law Review 487

⁹¹ *Ibid* at 487

⁹² Vlad Perju, ‘Impairment, Discrimination, and the Legal Construction of Disability in the European Union and the United States’ (2011) 44 Cornell International Law Journal 279, 310

The interpretation of the disability ground to encompass genetic discrimination would therefore be challenging based on the rationale of the *Chacon Navas* case. Two years later, the ECJ was again called upon to interpret the ground of disability, and this time the outcome was more favourable.

4.2.4 Coleman v Attridge Law

In the *Coleman v Attridge*⁹³ case, the ECJ held that the Framework Directive and in particular Articles 1, 2(1)(a) of the Directive, must be interpreted as meaning that the prohibition of direct discrimination in these provisions is not limited to people who are themselves disabled. The court held that “*where an employer treats an employee who is not disabled less favourably than another employee is, has been or would be treated in a comparable situation, and it is established that the less favourable treatment of that employee is based on the disability of his child whose care is primarily by that employee, such treatment is contrary to the prohibition on direct discrimination as provided for in Article 2(2)(a).*”⁹⁴ The court therefore extended protection under the Framework Directive to individuals who are not themselves disabled, but who are discriminated against ‘on the basis of disability.’ The decision is important, “*as it will prevent employers from treating those with disabled dependents less favourably than those with non- disabled dependents.*”⁹⁵

In extending protection to those individuals who are not themselves disabled, the focus is therefore taken away from the individual and placed on the discriminatory action. Pursuant to this case, the discriminatory use of genetic information may well be covered and prohibited under the Framework Directive. In circumstances where an employer makes an unfavourable decision based on an employee’s family history of illness, for example, where an employee’s mother had breast cancer or an employee’s father died of heart disease, these actions might be contrary to the Framework Directive, on the basis of the interpretation given in the *Coleman v Attridge* case. An employer might use an employee’s genetic history to make a decision in relation to hiring, promotion or potentially termination, even though the employee may not currently have a disability. The judgment in this case is also reflective of the social model of disability, in placing the focus on external factors, such as negative attitudes which operate to disable a person. However, it is noted that this case has attracted some criticism in terms of an over- extension of the concept of discrimination on the grounds of disability, and may have the effect of undermining the legal protection of persons with

⁹³ Case C-303/06 *S. Coleman v Attridge Law and Steve Law* [2008] ECR I- 05603

⁹⁴ *Ibid* at para 56

⁹⁵ Marguerite Bolger and Cliona Kimber, ‘Employment Equality’ in Maeve Regan (ed) *Employment Law* (Tottel Publishing 2009) 511

disabilities.⁹⁶ Nevertheless, it is a welcomed judgment in expanding the principle of non- discrimination in the EU.

4.2.5 Joined cases: Ring and Werge⁹⁷

A recent judgment of the Court of Justice of the European Union (CJEU) has further clarified the definition of disability. The joined case of *Ring and Werge* is particularly important in light of the fact that it is the first decision on the definition of disability under the Framework Directive since the EU concluded the CRPD in 2010. Importantly, the CJEU departed from the narrow approach taken in *Chacon Navas* and interpreted the concept of disability in light of Article 1 of the CRPD, which states: "*Persons with disabilities include those who have long- term physical, mental, intellectual or sensory impairments, which in interaction with various barriers may hinder their full and effective participation in society on an equal basis with others.*" The court therefore adopted the social model of disability in formulating a definition of disability.

In relation to the *Chacon Navas* case, it was acknowledged that the ruling was given before the entry into force of the CRPD.⁹⁸ In this regard, the court stated that "*the primacy of international agreements concluded by the European Union over instruments of secondary law means that those instruments must as far as possible be interpreted in a manner that is consistent with those agreements.*"⁹⁹ The decision acknowledged the position of the CRPD and its effect in the EU.

This ruling is certainly welcomed not only in terms of acknowledging the significance of the CRPD (and its conclusion by the EU), but also in constructing a definition of disability in line with the CRPD and reflective of the social model of disability. On consideration of whether the concept of disability in the EU encompasses genetic predisposition to disability and whether the Framework Directive reaches to include the protection of individuals with genetic predispositions to disability, this decision certainly supports such a positive interpretation.

4.2.6 Commission v Italy

Recently, the ECJ further clarified the current conception of disability in the case of *Commission v Italy*, and emphasised the relevance of the CRPD for the EU in

⁹⁶ Aart Hendriks, 'The UN Disability Convention and (Multiple) Discrimination: Should EU Non-Discrimination Law be Modelled Accordingly?' in Gerard Quinn and Lisa Waddington (eds) 2 *European Yearbook of Disability Law* (Intersentia 2010) 19, 20

⁹⁷ Joined cases C- 335/11 and C- 337/11 *HK Danmark, acting on behalf of Jette Ring v Dansk almennyttigt Boligselskab* (C-335/11) and *HK Danmark, acting on behalf of Lone Skouboe Werge v Dansk Arbejdsgiverforening, acting on behalf of Pro Display A/S, in liquidation* (C-337/11)

⁹⁸ *Ibid*, para 37

⁹⁹ *Ibid*, para 29

this area.¹⁰⁰ It confirmed that, pursuant to the CRPD, the definition of disability should be understood to refer to a limitation, resulting, *inter alia*, from a long term physical, mental or psychological impairment, which in interaction with various barriers may hinder a person's full and effective participation in employment on an equal basis with other employees.¹⁰¹ This case further clarifies the social model construction of disability in the EU and reflects the rationale of the CRPD.

4.2.7 Evaluation of the Framework Directive

The Framework Directive was a welcomed contribution to the EU human rights framework, and operates, *inter alia*, to advance the rights of persons with disabilities in employment. It also facilitates the inclusion of persons with disabilities in society. Although the Framework Directive was a landmark development in the protection of disability rights in employment, the concept of genetic discrimination is not specifically provided for in the legislation. By interpreting the 'disability' ground extensively, genetic discrimination could potentially be a covered ground of discrimination as discrimination on the basis of putative disability or assumed disability, or indeed on the basis of genetic predisposition to future disability. It can be assumed that 'genetic features' is encompassed within the ground of disability (as was averted to by Quinn in his examination of the Directive).

The case law of the European courts has also moved in a positive direction towards accepting such an interpretation. Although the *Chacon Navas* case was a restriction on the interpretation of the concept of disability at EU level, subsequent case law of the ECJ has been more favourable. This is evident particularly from the *Coleman* case, the *Ring and Werge* case and *Commission v Italy*. The interpretation of the disability ground has therefore evolved from a restrictive, medical model based definition, towards a wider, more holistic approach and one that is in line with the social construction of disability and the CRPD. It also creates the scope to encompass the grounds of genetic predisposition to disability and genetic information.

On evaluation of the Framework Directive generally in this area, it is acknowledged that it takes a broad approach to discrimination and provides for both direct discrimination and indirect discrimination. In certain circumstances, discrimination on the basis of one's genes may be construed as a form of indirect discrimination under the Framework Directive (as discussed). This may offer some degree of protection against those individuals, groups or races that suffer adverse treatment from an apparently neutral criterion, practice or provision. However, it is unlikely this would provide adequate or complete protection in this area.

¹⁰⁰ Case C-312/11 European *Commission v Italy* [2013] (4 July 2013)

¹⁰¹ Ibid

In addition, evaluation of the Framework Directive must refer to its limited scope in terms of applying solely in the employment context.¹⁰² It prohibits discrimination in the fields of employment, and vocational training, however it does not cover other fields in which genetic information might be misused, such as insurance. Therefore it is concluded that the Framework Directive does not offer complete and clear protection against discrimination on the grounds of genetic predisposition to disability or genetic information.

4.3 The Race Directive

This section will examine the Race Equality Directive as forming a key part of the EU non- discrimination and human rights framework, in implementing the principle of equal treatment between individuals irrespective of racial or ethnic origin and confirming the EU's commitment to fundamental human rights.¹⁰³ The Race Directive was adopted "*as part of a general effort of the European Union to boost its social legitimacy and its human face at a time of rising Euro- skepticism and xenophobia.*"¹⁰⁴ The focus on fundamental rights and freedoms also operates to enhance social and economic harmony in the EU.¹⁰⁵ The Race Directive has a reasonably broad scope of application and applies in both the employment context and in other contexts such as housing, education, health care, and social assistance, as well as in respect of other goods and services available to the general public.¹⁰⁶ The concept of discrimination is broadly drafted, and provides for both direct and indirect discrimination in a similar manner to the provisions in the Framework Directive.¹⁰⁷

The Race Directive has a number of unique elements and marks a welcomed contribution to the EU non- discrimination and human rights framework. The following sections will explore this Directive and highlight some of its intricacies. It has been observed that "*although the Race Equality Directive sits amidst a range of EU anti- discrimination legislation, it possesses a number of features that have underscored its relative strength.*"¹⁰⁸ Bell highlights some of these unique

¹⁰² Anna Lawson, 'The UN Convention on the Rights of Persons with Disabilities and European Disability Law: A Catalyst for Cohesion?' in Oddný Mjöll Arnardóttir and Gerard Quinn (eds) *UN Convention on the Rights of Persons with Disabilities* (Martinus Nijhoff Publishers 2009) 105

¹⁰³ The Race Directive

¹⁰⁴ Grainne de Burca, 'The Trajectories of European and American Antidiscrimination Law' (2012) 60 *American Journal of Comparative Law* 1, 10. See also Adam Tyson, 'The Negotiation of the European Community Directive on Racial Discrimination' (2001) 3 *European Journal of Migration and Law* 199, and more generally Mark Bell, *Anti- Discrimination Law and the European Union* (Oxford University Press 2002)

¹⁰⁵ See Catherine Barnard, *EC Employment Law* (3rd edn, Oxford University Press 2006) 310

¹⁰⁶ Phillipa Watson, 'Equality Between Europe's Citizens: Where does the Union now Stand?' (2011-2012) 35 *Fordham International Law Journal* 1426, 1460

¹⁰⁷ Article 1 and Article 2(2)(a) and (b)

¹⁰⁸ Mark Bell, 'EU anti- racism policy: the leader of the pack?' in Helen Meenan (ed) *Equality Law in an Enlarged European Union – Understanding the Article 13 Directives* (Cambridge University Press 2007) 181

features. First, in comparison to the Framework Directive, the Race Directive's scope of application is reasonably broad. As noted, it applies in both the employment context and in other contexts such as housing, education, health care, and social assistance, as well as in respect of other goods and services available to the public. Another unique element of the Race Directive, in comparison to the other 'Article 13 Directives' is the fact that it contains relatively few exceptions to the principle of equal treatment, thereby enhancing the effectiveness of the protections offered.¹⁰⁹ Accordingly, there are reasonably robust protections under the Race Equality Directive, with regard to race and ethnicity.

Another interesting observation is the decision to segregate race in an isolated Directive. It is noted that there was a great deal of political support and public backing that prompted the recognition that this form of discrimination deserved individually tailored protection, in the form of a specifically tailored Directive. In this regard, it is noted that "*although this pragmatism paid dividends in the form of a stronger and broader Directive, its legacy is the legal stratification of race and ethnicity as separate from the other discrimination grounds.*"¹¹⁰ There are arguments for and against such a mode of regulation.

This approach has been criticised as being impractical and not truly reflective of the reality of discrimination, which often cannot be separated according to particular categories.¹¹¹ Bell makes two points in support of this submission. He points out the confusion and lack of clarity surrounding the meaning of 'racial or ethnic origin.'¹¹² For example, the connection between race and religion illustrates the dilemmas encountered in differentiating grounds of discrimination in the way envisaged by the Directives. These two grounds are often interconnected, particularly in certain jurisdictions, thereby making it challenging to separate these grounds. In addition, certain discriminatory grounds can combine to constitute particularly undesirable discrimination and adverse treatment.¹¹³ For example, evidence has shown that certain types of third country national women have significantly reduced employment rates than those of either third country national men or indeed women in general.¹¹⁴

However, the merits of this approach can equally be argued. Singling out race, or any other discriminatory ground, in an individual Directive has a certain expressive value that is welcomed in this area. Following an evident history of discrimination on the grounds of race or ethnicity in the EU, there was an evident need for a strong framework of protection on these grounds. In addition, such

¹⁰⁹ Ibid

¹¹⁰ Ibid at 183

¹¹¹ Ibid at 183

¹¹² Ibid at 183

¹¹³ Ibid at 184

¹¹⁴ Commission, 'Employment in Europe 2003' (Luxembourg, Office for the Official Publications of the European Communities) 198

discrimination stretches across all facets of life, from employment, education and goods and services. Therefore, discrimination on these grounds may adversely affect an individual's access to a range of social goods and services, as well as an individual's active participation in society. In light of the potential for stigma, segregation and marginalisation, there are strong societal and public policy reasons in favour of such a legislative instrument.

4.3.1 Evaluation of the Race Directive

The Race Directive forms an important part of EU non- discrimination law and EU human rights law in general. It offers broad protection addressing discrimination on the grounds of race or ethnic origin across a broad spectrum. In the current debate, it is observed that certain genetic conditions are more prevalent in certain races and ethnic minorities, and therefore genetic discrimination could possibly be provided for as a form of indirect discrimination, where, for example, a certain practice or provision puts persons of a particular race or ethnicity at a particular disadvantage compared with other persons or groups. Indeed, historic incidences of discrimination in the US and Europe illustrate the potential for this insidious discrimination to occur. As noted, in the 1970s in the US, a genetic screening programme which was introduced with the intention of identifying those susceptible to sickle cell anaemia had the effect of discriminating against African-Americans and highlighting existing racial inequalities.

4.4 The Gender Equal Treatment Directive

This section will examine the Gender Equal Treatment Directive, another 'Article 13 Directive' that forms a key part of the EU non- discrimination framework. In 2004 a Directive was introduced, which extends the principle of equal treatment between men and women to access to and supply of goods and services in the EU.¹¹⁵ It provides a common framework throughout the EU for the prohibition of discrimination based on gender in the access to and supply of goods and services.¹¹⁶ The principle of equal treatment under this Directive encompasses a prohibition on both direct and indirect discrimination.¹¹⁷ The Directive has a strong human rights ethos. In this regard, it is particularly interesting to note that Recital 4 to the Directive makes express reference to Articles 21 and 23 of the CFREU, which prohibits any discrimination based on gender and requires that equality between men and women is ensured in all areas.¹¹⁸ This is important in further reinforcing the relevance of the CFREU as a core element of the EU non-discrimination and human rights framework.

¹¹⁵ The Equal Treatment Directive

¹¹⁶ Ibid, Article 1

¹¹⁷ Ibid, Article 4(1)(a) and (b)

¹¹⁸ CFREU Article 23

It is relevant to highlight that this Directive has specific provisions in relation to insurance and use of gender for actuarial purposes. Article 5(1) provides “*Member States shall ensure that, in all new contracts concluded after 21 December 2007, the use of sex as a factor in the calculation of premiums and benefits for the purposes of insurance and related financial services shall not result in differences in individuals’ premiums and benefits.*”¹¹⁹ This provision is however narrowed and restricted by Article 5(2), which provides that Member States could decide to allow proportionate differences in “*premiums and benefits where the use of sex is a determining factor in the assessment of risk, based on relevant and accurate actuarial and statistical data.*”¹²⁰ However, Article 5(2) has been challenged and has been found to be unlawful by the ECJ.

4.4.1 Test- Achats case

In consideration of Article 5(2), a landmark case was handed down by the ECJ, known as “*Test Achats*.”¹²¹ In this case, the ECJ held Article 5(2) to be invalid. Specifically, the court found that the possibility of the Member States maintaining an exemption from the requirement in Article 5(1) of unisex premiums and benefits, was in violation of the fundamental principle of equal treatment, and accordingly is to be regarded as invalid from 21 December, 2012.¹²² Therefore, the court found that the derogation from the principle of equal treatment in the insurance sector, pursuant to Article 5(2), was unlawful.¹²³

As noted, the CFREU has an important position in the EU legal framework, and therefore is a key source of reference for the ECJ. The key provisions referred to in this case are Article 21 and Article 23 of the Charter. Article 21 of the CFREU prohibits any discrimination based, *inter alia*, on sex.¹²⁴ In addition, this provision is complemented by Article 23 of the CFREU, which requires equality between women and men.¹²⁵ The case therefore illustrates the prime position of fundamental human rights within the EU legal order.¹²⁶

Following the *Test- Achats* case, the Commission issued a Communication in December 2011, offering guidelines with the objective of ensuring compliance by

¹¹⁹ Equal Treatment Directive Article 5(1)

¹²⁰ Equal Treatment Directive Article 5(2)

¹²¹ *Association Belge des Consommateurs Test- Achats ASBL v Conseil des ministres*, Case C- 236/09 [2011]

¹²² Phillipa Watson, ‘Equality Between Europe’s Citizens: Where does the Union now Stand?’ (2011-2012) 35 Fordham International Law Journal 1426, 1458- 1459

¹²³ *Ibid* at 1480

¹²⁴ CFREU Article 21

¹²⁵ CFREU Article 23

¹²⁶ Felipe Temming, ‘Case Note- Judgment of the European Court of Justice (Grand Chamber) of 1 March 2010: ECJ finally paves the way for unisex premiums and benefits in insurance and related financial service contracts’ (2012) 13 German Law Journal 1 106, 110

insurance companies with this decision.¹²⁷ However, these guidelines are non-binding and the Commission's position is "*without prejudice of any interpretation the Court of Justice may give to Article 5 in the future.*"¹²⁸ It is observed that although an amendment to this Directive is feasible from a legal perspective, it is questionable whether this is likely to happen from a political perspective.¹²⁹

4.4.2 Implications of the *Test- Achats* case

The *Test- Achats* case was a landmark ruling and has been described as "*a milestone in combating discrimination based on sex.*"¹³⁰ Setting different premiums and benefits for men and women in private insurance is no longer permitted. It is pointed out that the ECJ was ambiguous to a certain degree, in relation to the impact of its ruling on "*current life insurance contracts, the effect on occupational pensions and the use of age and disability as a differentiation factor in insurance contracts.*"¹³¹ This case has relevance not only in respect of the use of gender as a risk factor in private insurance, but also potentially in respect of using other insurance risk factors. It is pointed out that the ECJ's ruling may be significant as regards the use of further factors such as age, disability and perhaps genetic information. Indeed the decision may pave the way for further equality and acknowledgment of fundamental rights in the formation of private insurance contracts. It may trigger a need to re- conceptualise the traditional principles of insurance law.

4.4.3 Evaluation of the Gender Equal Treatment Directive

The Gender Equal Treatment Directive is an important part of the EU non-discrimination package. It broadly proscribes both direct and indirect discrimination on gender grounds in the area of goods and services. It also highlights the application of non- discrimination law to insurance. Further, the interpretation of these provisions in the *Test Achats* case is a positive development in non- discrimination law, and may have the effect of expanding the application of non- discrimination law in the area of insurance, and it arguably

¹²⁷ Commission Communication to Member States on Guidelines on the Application of Council Directive 2004/113/EC to Insurance, in the Light of the Judgment of the Court of Justice of the European Union in Case C- 236/09 (*Test- Achats*) 2011 OJ L 9497

¹²⁸ *Ibid*, para 4

¹²⁹ Geert De Baere and Eveline Goessens, 'Gender Differentiation in Insurance Contracts after the Judgment in Case C- 236/09, Association Belge des Consommateurs *Test- Achats* ASBL v Conseil Des Ministres' (2011- 2012) 18 *Columbia of European Law* 339, 353. De Baere and Goessens note that the Commission, which holds the legislative initiative, appears to have no intention to modify Article 5 of the Directive

¹³⁰ Felipe Temming, 'Case Note- Judgment of the European Court of Justice (Grand Chamber) of 1 March 2010: ECJ finally paves the way for unisex premiums and benefits in insurance and related financial service contracts' (2012) 13 *German Law Journal* 1 106, 112

¹³¹ Geert De Baere and Eveline Goessens, 'Gender Differentiation in Insurance Contracts after the Judgment in Case C- 236/09, Association Belge des Consommateurs *Test- Achats* ASBL v Conseil Des Ministres' (2011- 2012) 18 *Columbia of European Law* 339, 352

supports the argument in favour of non- discrimination on the grounds of genetic information in insurance.

4.5 Proposals for a new non – discrimination Directive

This section will briefly highlight proposals for a new non- discrimination framework in the EU that have taken place in the last few years. Although the proposed draft Directive failed to reach consensus and faced much opposition, it is important to highlight the potential scope of EU non- discrimination law and the implications this may have for the EU. This section will also examine the potential implications that this proposed legislative endeavour might have on the use of genetic information in the EU, from a non- discrimination perspective.

In 2008 the Commission made proposals for a new non- discrimination Directive,¹³² with the objective of expanding the scope of non- discrimination law in the EU. These proposals were for a new Directive to address discrimination on the basis of sexual orientation, religion, disability and age, which aimed to extend the existing provisions in place for racial and sex discrimination to these other grounds. The proposal seeks to extend the EU's non-discrimination legislation beyond the realm of employment and occupation, with the proposed Directive addressing discrimination in the fields of social protection, social advantages, education, and access to and supply of public goods and services. However, the proposed Directive was strongly opposed, and the Member States have failed to reach consensus.

The proposed Directive would have particular implications in the area of disability and would operate to enhance the rights of persons with disabilities in the EU. The proposal provides that a failure to provide reasonable accommodation (for persons with disabilities) is a form of discrimination.¹³³ This is in line with the CRPD, which expressly treats such a failure as a form of discrimination.¹³⁴ These strong provisions providing for reasonable accommodation would certainly be welcomed in terms of bringing EU non- discrimination law in line with the CRPD.

It is submitted that the proposal would also have potential implications for the use of genetic information in the EU. It is observed that insurance companies are covered under the scope of the draft Directive. The proposal permits proportionate differences in treatment in the provision of financial services where, for the product in question, the use of age or disability is a key factor in the assessment of risk based on relevant and accurate actuarial or statistical data.¹³⁵ In theory, insurance companies are obliged to comply with these provisions, under the proposed Directive, however the effectiveness of these provisions is

¹³² Proposal for a Council Directive on implementing the principle of equal treatment between persons irrespective of religion or belief, disability, age or sexual orientation, COM (2008) 426 final

¹³³ Ibid, Article 2(5)

¹³⁴ Ibid, Article 4(1)(a)

¹³⁵ Ibid, Article 2(7)

diluted to a certain extent by this exception in respect of using age or disability in the assessment of risk. Although this exception might appear to permit the use of age or disability, as regards the use genetic information obtained from other sources, this information may not be based on relevant and accurate actuarial or statistical data, in light of the often inaccurate and limited predictive value of most genetic tests, and genetic information from other sources.

4.5.1 Opposition to the draft Directive

The proposed Directive is a welcomed addition to the EU non- discrimination framework in expanding the scope of protections. It would greatly expand the scope of EU non- discrimination law to cover discrimination in a range of third party contexts. However, this proposed Directive has encountered strong opposition and after several years of uncertain negotiations, it does not appear likely that the proposal will be adopted. In this context, de Burca observes that the “*political enthusiasm for the enactment of antidiscrimination measures is waning at the EU level.*”¹³⁶ She also notes the “*significant pushback and resistance from political and societal forces within European states.*”¹³⁷ It is submitted that the recent economic crisis in the EU has had an impact upon political and societal priorities, with a focus on economic objectives. In this regard, it is also noted that a current priority of the EU in this area is the Accessibility Act, which focuses to a large extent on the internal market and facilitating the economic objectives of the EU. Therefore, although an admirable legislative endeavour, we cannot firmly rely upon these draft proposals as representative of the future orientation of EU non- discrimination law.

5. Other relevant documents in the EU

This section will highlight key policy and other documents of the EU in this area.

In 1989 the European Parliament adopted a Resolution on the ethical and legal problems of genetic engineering.¹³⁸ The Resolution provides, *inter alia*, a prohibition on the use of genetic testing by insurers. It also contains provisions on employment, and calls for, *inter alia*, “*a statutory ban on the selection of workers on the basis of genetic criteria.*”¹³⁹ This Resolution is evidence of a certain EU level consensus on this issue and an early indication of a need to regulate this

¹³⁶ Grainne de Burca, ‘The Trajectories of European and American Antidiscrimination Law’ (2012) 60 American Journal of Comparative Law 1, 21. See also Phillipa Watson, ‘Equality Between Europe’s Citizens: Where does the Union now Stand?’ (2011- 2012) 35 Fordham International Law Journal 1426, 1484

¹³⁷ Ibid

¹³⁸ Parliament Resolution of 17 April 1989 on the ethical and legal problems of genetic engineering [1989] OJ C 96/165- 171

¹³⁹ Ibid

area, however, it is noted that there have been no concrete legislative developments since.

In 2004 the European Commission released a document containing 25 recommendations on the Ethical, Legal and Social Implications of Genetic Testing.¹⁴⁰ The recommendations cover extensive ground from the area of regulation, to the quality of genetic testing services, as well as the ethical, social and legal issues arising. In terms of the general framework, the first recommendation advocated the need for universal standard definitions (for genetic testing and genetic data), to give clarity and certainty to this area.¹⁴¹

The document contains a specific recommendation on the need for privacy, confidentiality and autonomy.¹⁴² Arguably, these recommendations are fulfilled in the current data protection framework, and particularly with the recent proposed reform of data protection law. The document also contains a specific recommendation indicating the need for protection from discrimination. In this regard, it recommends that EU- level regulations that address discrimination issues should be promoted.¹⁴³ As noted above in the discussion of the data protection position, this document made recommendations in favour of the equal protection of all medical data (thereby avoiding reliance on the notion of genetic exceptionalism).¹⁴⁴ The expert group also recognised the need for public engagement with these issues. To this end, it recommends public awareness raising and education, as well as a multidisciplinary public dialogue on these issues.¹⁴⁵

Although non- binding, this document is a core policy initiative in this area at EU level, and acts as a guidance tool for any legislative and policy endeavours in the area. It takes an interdisciplinary approach towards identifying the key ethical and legal considerations arising in this area, and acknowledging the interested stakeholders who should contribute to this debate. This document also recognised gaps in protection as regards existing law, for example, with reference to the recommendation for EU- level regulation to address discrimination in this area. It is observed that since the publication of this document, there has not been any firm action or clear legislative endeavours in this area at EU level.

¹⁴⁰ European Commission 25 Recommendations on the Ethical, Legal and Social Implications of Genetic Testing, (European Commission Expert Group, 2004). Available at: http://ec.europa.eu/research/conferences/2004/genetic/pdf/recommendations_en.pdf (assessed 31st January 2013)

¹⁴¹ Ibid, Recommendation 1

¹⁴² Ibid, Recommendation 10

¹⁴³ Ibid, Recommendation 11

¹⁴⁴ Ibid, Recommendation 3

¹⁴⁵ Ibid, Recommendations 4 and 5

It is also relevant to note some of the provisions of the European Ombudsman's Code of Good Administrative Behaviour.¹⁴⁶ Article 5 relates to non- discrimination and prohibits, *inter alia*, "...any unjustified discrimination between members of the public based on... genetic features... disability ..."¹⁴⁷ The language of this provision is reflective of the CFREU, and further highlights the intention at EU level to outlaw discrimination on the basis of 'genetic features' and other grounds. In addition, a preliminary examination of the cases from the European Ombudsman revealed that there have been no cases decided on the issues of genetic discrimination and genetic privacy. However, this does not discount the possibility of similar cases involving the EU institutions in the future.

The above documents and policy endeavours are an important indication of an EU level awareness of the need to address the area of advancing genetic science and the issues that arise. The presence of these documents can perhaps nudge the EU to revisit this area and seriously reflect upon the need for EU level action in this area.

6. Conclusion

This chapter has provided a comprehensive overview of the current EU framework relevant in the area of non- discrimination and data protection. As evident in part one of this chapter, human rights form a key part of the EU legal order. The incorporation of the CFREU as a binding legal instrument, with the same value as the treaties is a significant development in this area, and it is submitted that this development may provoke EU action and re- orient existing EU rights and principles. In this regard, it is clear that the interpretation of human rights in the EU is evolving, for example, as is illustrated by the inclusion of 'genetic features' in the CFREU. It also highlights the increasing close connection between the Council of Europe and the EU, particularly as many of the provisions of the CFREU are reflected in the ECHR. In addition, the future accession of the EU to the ECHR is further illustration of the growing harmony between the two entities.

This chapter submits that current EU data protection law offers reasonably strong privacy protections, with the objective of protecting an individual's personal data. It is asserted that the data reform process will greatly enhance the protection of genetic privacy. Such a regime will have the effect of protecting the access to and disclosure of genetic data. By safeguarding genetic data in this manner, an individual's right to privacy will be respected. This provokes the question as to whether strong data protection laws in the EU are sufficient to control genetic information and protect against misuse. As concluded in chapter 5, it is doubtful

¹⁴⁶ See <http://www.ombudsman.europa.eu/en/resources/code.faces#/page/1> (accessed 7 July 2013)

¹⁴⁷ European Ombudsman's Code of Good Administrative Behaviour, Article 5(3)

that such a framework would fully protect individuals in this regard. However, these developments are a key legislative starting point in this area.

This chapter highlights the strong non-discrimination framework in the EU, which aims to protect against discrimination on a range of grounds, and in a variety of contexts. The non-discrimination regime in the EU is framed around a number of Directives. As illustrated above, the EU takes a broad approach to the concept of discrimination, by encompassing both direct and indirect discrimination. In the context of this debate, this is welcomed. It is submitted that discrimination on the basis of one's genetic information can occur through both overt and covert means, and therefore such comprehensive protections are appealing.

Although there is no explicit provision for discrimination on the grounds of genetic predisposition to disability under the Framework Directive, the scope of the disability ground has been positively interpreted to encompass a broad definition of disability, in line with the CRPD. The case law of the ECJ in this area has evolved to reflect current thinking and the social construction of disability. Therefore, it is likely that discrimination on the basis of genetic predisposition to disability is protected under the Framework Directive. However, from a non-discrimination perspective, these protections do not stretch further than the employment context, and accordingly a gap in the law can be identified.

The Gender Equal Treatment Directive and the proposed draft non-discrimination Directive offer interesting insights into the non-discrimination framework in the EU, particularly in the insurance field. It is also acknowledged that the Race Directive serves as an example of singling out a particular discriminatory ground in an individually tailored directive. However, there is incomplete protection for genetic information at EU level. The gaps identified in the non-discrimination framework would point towards the need for further protections to comprehensively cover discrimination on the basis of genetic predisposition to disability in the areas of employment and insurance.

On evaluation of the EU position in this area, it is clear that there is currently no concrete legal framework at EU level to address the discriminatory use of genetic information or to protect genetic privacy. Although the strong data protection regime is a good starting point, the gaps identified in the non-discrimination framework would point towards the need for further protections to comprehensively cover discrimination on the basis of genetic predisposition to disability in the areas of employment and insurance. In light of these submissions, the following chapter will further establish and build a case for EU action in this area. In particular, it will highlight the evident patchwork of protections amongst the EU Member States, and will examine the EU competence to act in this area.

PART 4: BUILDING THE CASE FOR A EUROPEAN UNION REGULATORY FRAMEWORK

Part one of this thesis highlighted and explored the legal and ethical issues arising in light of advancing genetic science and technology. Against a disability approach to this issue, part two explored the imperative for regulation and the choice of regulatory frameworks, as well as highlighting the comparative law benchmarks. Part three gave an overview of the European position, both at Council of Europe level, and EU level, and illustrated some of the apparent gaps in protection. Having elucidated the key issues arising, highlighted the need for appropriate regulation, and having identified the gaps in the current regulatory regime, this part will build a case for EU level action in this area. The final part of this thesis comprises two chapters: chapter 10 entitled 'A case for EU action' and finally chapter 11, the recommendations and conclusions.

Chapter 10: A case for European Union level action

1. Introduction

This chapter will highlight the merits and challenges of EU level action to regulate the use of genetic information. On considering the need for an EU level response, reference is made to the speed at which genetic science is advancing and the subsequent potential for misuse and violation of fundamental human rights, as discussed in Part 1. The relevant international and comparative benchmarks and the current gaps in EU level protection are also acknowledged, and compound the need for a new regulatory regime in this area.

In building a case for EU level action, this chapter will firstly examine some of the individual EU Member State responses in this area, and highlight the diversity of approaches. This section will illustrate the different regulatory approaches taken and the varying levels of protection offered by these different models, resulting in a potentially unworkable patchwork of protection. This chapter will argue that this disparity at EU Member State level is not a suitable or appropriate means of regulation. It will argue that a preferable regulatory framework is a comprehensive, uniform one at EU level. This would operate to create clarity and consistency in this area. It would also enhance the functioning of the internal market, as companies operating across borders would not have the administrative, legal and other burdens of complying with different regulatory systems.

A further argument in support of an EU level response is based on the rationale that preemptive legislation is necessary in order to enhance and maintain public confidence in genetic science and technology in the EU. It is submitted that the practical application of genetic science and further scientific innovation may be stifled if individuals are fearful of such developments and unwilling to engage in genetic testing. This argument is also in line with economic objectives, particularly as science and technology are key drivers of economic success in the EU.¹ On building a case for EU action in this area, this chapter will also discuss the evidence (or lack thereof) of misuse of genetic information in the EU and other jurisdictions, with a view to anticipating future misuse. On proposing EU level legislation, the second part of this chapter will consider the legal bases for action at EU level, with a focus on the EU's competence in the area of non-discrimination and the EU's competence to facilitate and maintain the internal market. In support of an EU level response, this section notes the importance of the CRPD and the CFREU.

¹ Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions: Horizon 2020 – The Framework Programme for Research and Innovation COM (2011) 808 final

This chapter will look at the potential challenges to action in this area. This includes challenges that may be faced as regards the competence of the EU in this area. It also includes evidentiary issues that need to be considered. The chapter will then explore the shape that the proposed EU level approach might take, and the mode of regulation. It will also address the scope and content of the proposed legislation. Finally, this chapter will consider whether legislation alone is enough to comprehensively address this area.

2. Diversity of approaches in the EU Member States

Although a comprehensive analysis of the position in every EU Member State is outside the scope of this thesis, the objective of this section is to provide an overview of some individual approaches in the EU. A broad overview will be given and then selected examples will be examined in a general fashion. The aim is to illustrate the varying laws that exist, with the result of creating a patchwork of varying protections. It will therefore illustrate that there does not appear to be any harmony amongst the MS as regards the scope of protections, or the manner in which genetic information is protected.²

Some EU countries have enacted genetic specific legislation in this area, for example, Germany,³ Sweden,⁴ Austria,⁵ Portugal,⁶ Latvia,⁷ Estonia,⁸ and Czech Republic.⁹ In contrast, other EU countries, including Ireland, Belgium, Spain, Bulgaria and the United Kingdom have not enacted specific legislation to address genetic information, but have regulated the area under other regulatory frameworks. For example, Belgium was the first EU country to introduce an insurance law with a prohibition on the use of genetic information for underwriting purposes.¹⁰ The Netherlands has taken the approach of regulating the use of all health- related information.¹¹ In Bulgaria, the area is also regulated under general health legislation,¹² as is Denmark.¹³ Similarly, in Cyprus¹⁴ and Hungary,¹⁵ the area is covered by health care legislation. In Ireland, genetic information is

² Sirpa Soina, 'Genetic Testing legislation in Western Europe – a fluctuating regulatory target', (2012)

³ Journal of Community Genetics 143. See also Orsolya Varga et al, 'Definitions of genetic testing in European legal documents' (2012) 3 Journal of Community Genetics 125

³ Human Genetic Examination Act (Genetic Diagnosis Act GenDG) 374/09, 2009

⁴ Genetic Integrity Act No. 2006: 351

⁵ Gene Technology Act-GTG (Gentechnikgesetzes BGBl 510/1994)

⁶ Personal Genetic Information and Health Information Act, Law No. 12/2005 of 26 January 2005

⁷ Human Genome Research Law of 13 June 2002

⁸ Human Genes Research Act, of 13 December 2000 (RT I 2000, 104, 685)

⁹ Act No. 373/ 2011 Sb. on Specific Health Services

¹⁰ Law of 25 June 1992 on terrestrial insurance contracts, Article 95

¹¹ Law on Medical Examination of 1997 (1 January 1998)

¹² Bulgarian Health Care Reform and Health Care Act 2004 (Bulgarian Health Act 2005) SG []

¹³ Danish Act on the use of health on the labour market (1996)

¹⁴ The Safeguarding and Protection of Patients' Rights Law 2004, 1 of 2005

¹⁵ Health Care Act of 1997 CLIV

regulated under disability rights legislation¹⁶ and in Finland, the area has been addressed under employment privacy legislation.¹⁷

Spain has regulated the area under general biomedical legislation.¹⁸ France has included protection of genetic information under its bioethical legislation.¹⁹ The legislation in Slovakia focuses mainly in the criminal justice context.²⁰ In Greece, it amended its Constitution to provide for the privacy of genetic information.²¹ Highlighting a different mode of regulation, the UK has adopted a moratorium in the insurance industry.²²

It is further noted that other European countries (outside of the EU) have also enacted genetic specific legislation, such as Switzerland.²³ It is also observed that Norway (outside of the EU) has included protection in this area under biomedical legislation.²⁴

In addition, different regulatory frameworks can be identified in these different jurisdictions. For example, Austria, Ireland, Spain and the Netherlands take an evident privacy or data protection approach, whereas Germany takes an evident non- discrimination approach. The Portuguese approach appears to be a combination of the non- discrimination and data protection models. In Sweden, there is an evident human rights based approach, with a focus on the dignity and integrity of the person.

Based on a preliminary examination, it is also observed that some EU Member States do not appear to have legislated in this area, either through genetic specific laws or through other laws. For example, Slovenia, Lithuania, Luxembourg, Malta, Italy, Poland and Romania. There have however been some soft law endeavours in these countries. For example, in Italy, the Genetic Authorization for the Processing of Genetic Data was introduced in 2011.²⁵ In Poland, Article 29 of the Code of Medical Ethics covers the protection of genetic information, and provides for the confidentiality of such information.²⁶

In addition, in the individual Member States the area of genetic testing and regulation of genetic information may be dealt with under other legal frameworks,

¹⁶ Disability Act 2005 Act No 14 of 2005

¹⁷ Act on the Protection of Privacy in Working Life (759/2004)

¹⁸ Law 14/2007 of 3 July on Biomedical Investigations

¹⁹ Bioethical Act 2004- 800 that modified the civil code and public health code. (Revisions by acts 2011- 267 and 2011- 814)

²⁰ Act No 417/ 2002 Use of DNA analysis for identification of persons

²¹ Constitution of Greece, Article 5, para 5

²² Concordat and Moratorium on Genetics and Insurance

²³ Federal Act on Human Genetic Analysis, Act of 8 October 2004

²⁴ Act of 5 December 2003 No. 100 relating to the application of biotechnology in human medicine

²⁵ Genetic Authorization for the Processing of Genetic Data (Italian Data Protection Authority 2011). See also Linda Battistuzzi, 'Definitions of genetic testing in Italian Legal Documents' (2013) 4 Journal of Community Genetics 289

²⁶ Code of Medical Ethics, adopted by the General Medical Assembly in 1991

such as equality and non- discrimination law and data protection law. A comprehensive analysis of these frameworks is outside the scope of this thesis, however, the main objective of the analysis in this section is to highlight the diversity in the levels of protection in this area at Member State level.

A comprehensive analysis of national legislative frameworks is outside the scope of this thesis, however, the following section will highlight the diversity in the levels of protection in the Member States. It will examine the following regulatory frameworks in greater detail: Germany, Austria, Portugal, Sweden, the Netherlands, Ireland and the UK. The rationale for examining the position in Germany, Austria, Portugal and Sweden is that these present clear examples of genetic specific, stand- alone laws which have been introduced. This thesis looks at the position in the Netherlands and Ireland, to illustrate a further approach taken. Finally, this section will look at the regulatory position in the UK. The decision was made to look at the UK model of the moratorium, as this is regarded as a successful example of a soft law approach in this area.

2.1 Genetic specific laws

These genetic specific laws noted above are reflective of the concept of genetic exceptionalism, by singling genetic information out in stand- alone legislation. This section will give a brief overview of some of the relevant laws adopted in these countries.

2.1.1 Germany

In 2010 the Human Genetic Examination Act, a federal law, was introduced in Germany.²⁷ It aims to prohibit discrimination based upon genetic characteristics. It focuses on the duty to protect human dignity and to ensure the right to self-determination for all individuals.²⁸

The scope of application of the legislation is broad and applies, *inter alia*, in the employment and insurance contexts. The German framework takes primarily a non- discrimination approach towards regulating genetic information. Article 4 contains a general non- discrimination provision. Article 18 prohibits the use of genetic information in the insurance context,²⁹ and contains relatively strong provisions against the use of genetic test results by insurers. Article 19 applies to genetic testing in employment and prevents an employer demanding genetic test results either before or during employment.³⁰ In addition, Article 21 provides a broad provision against discrimination on the basis of genetic characteristics in

²⁷ Human Genetic Examination Act (Genetic Diagnosis Act GenDG) 374/09, 2009

²⁸ Ibid, Article 1

²⁹ Ibid, Article 18

³⁰ Ibid, Article 19

the employment context.³¹ These provisions offer a relatively high level of protection against the misuse of genetic information in employment. Article 20 attempts to strike a balance and relates to genetic testing for occupational safety (setting out a number of conditions necessary in order to conduct genetic testing in these circumstances).³²

The legislation also has implications for Direct to Consumer (DTC) genetic testing, with the Act restricting such services. Genetic tests can only be carried out by a licensed doctor and with the patient's consent.³³ Arguably, the Act restricts the ability of individuals to access their own genetic information directly, with potential implications for the right to exercise one's autonomy.

Germany adopts a very protective and cautious approach towards this area, which is perhaps reflective of the objectionable history of eugenics and abuse of genetic science which took place in Germany. It is an example of a genetic specific model which takes primarily a non-discrimination approach towards regulating genetic information.

2.1.2 Austria

The Austrian Gene Technology Act³⁴ was adopted in 1995, and an amendment on Gene Testing and Gene Therapy on humans (section IV) came into force in 2005. Article 65 limits genetic testing for medical purposes only.³⁵ Similar to the German legislation, the Austrian legislation also applies in the employment and insurance context. However, the Austrian legislation takes primarily a privacy and data protection approach to this area.³⁶ Article 67 imposes a prohibition on the collection and use of data from genetic tests for certain purposes.

Pursuant to Article 67, employers and insurers are prohibited from collecting, demanding, accepting or using results of genetic tests from employees, job applicants or insureds.³⁷ The legislation provides requirements of informed consent, qualification of the medical professionals involved, and compulsory registers of the institutions providing services. It is observed that *"the legislation regulates genetic activities in a rather strict and detailed manner,"*³⁸ and highlights the operation of a data protection approach to regulating genetic information.

³¹ Ibid, Article 21

³² Ibid, Article 20 (3)

³³ Ibid, Article 8

³⁴ Gene Technology Act-GTG (Gentechnikgesetzes BGBl 510/1994)

³⁵ Ibid, Article 65

³⁶ Ibid, Article 71

³⁷ Ibid, Article 67

³⁸ Sirpa Soina, 'Genetic Testing legislation in Western Europe – a fluctuating regulatory target' (2012) 3 Journal of Community Genetics 143, 147

2.1.3 Sweden

Sweden introduced the Genetic Integrity Act in 2006.³⁹ It is broadly drafted legislation, which governs the use of a variety of genetic technologies, including genetic testing, prenatal and preimplantation genetic diagnosis. In reflecting a human rights approach, the objective of the Act is to safeguard a person's integrity.⁴⁰ In maintaining a focus on the integrity of the person, this law reflects the proposition that genetic make-up, to a certain degree, determines the uniqueness and identity of a human being.⁴¹ The Swedish legislation maintains a strong emphasis on the ethical issues arising from advancing genetic technology. The legislation contains specific provisions for use of genetic information by insurers, which is permitted (albeit under certain conditions).⁴² There are no specific provisions in the legislation for use of genetic information by employers. However, there is a very general prohibition against using genetic information in connection with an agreement between two parties.⁴³ Therefore, presumably the legislation also applies in the employment relationship context. The Swedish law takes a relatively broad approach, from a rights based perspective.

2.1.4 Portugal

Portugal introduced the Personal Genetic Information and Health Information Act in 2005.⁴⁴ The law has a broad scope and regulates the use of both genetic information and general health information. It also sets out principles governing, *inter alia*, genetic testing, genetic databases and biobanks, and the conduct of research. There are specific provisions for employment and insurance. The legislation provides that insurance companies are not permitted to seek genetic information⁴⁵ and employers can only use this information for certain health and safety purposes.⁴⁶ In terms of regulatory approach, the legislation contains a general non-discrimination provision.⁴⁷ There is also an evident privacy approach, and it contains several references to confidentiality and data protection.⁴⁸

2.2 Other approaches

The following sections will look at other approaches that have been taken in the EU.

³⁹ Genetic Integrity Act No. 2006: 351

⁴⁰ Chapter 1, s 1

⁴¹ Sirpa Soina, 'Genetic Testing legislation in Western Europe – a fluctuating regulatory target' (2012) 3 Journal of Community Genetics 143, 149

⁴² Chapter 2, s 2

⁴³ Chapter 2, s 1

⁴⁴ Personal Genetic Information and Health Information Act, Law No. 12/2005 of 26 January 2005

⁴⁵ Ibid, Article 12

⁴⁶ Ibid, Article 13

⁴⁷ Ibid, Article 11

⁴⁸ Ibid, Article 4, Article 6

2.2.5 The Netherlands

In comparison to some of the genetic specific laws as illustrated above, a more general approach has been taken in the Netherlands, where the regulatory response has been relatively broad ranging. The Medical Examination Act 1997 seeks to regulate the use of medical examinations and to restrict the scope of health inquiries, therefore taking a broad approach encompassing health information generally. The legislation applies in both the employment and insurance contexts. The legislation imposes restrictions on employers and insurers as regards requesting genetic tests and using test results.⁴⁹ In adopting primarily a privacy approach, Article 3 provides that a medical test must not cause an excessive invasion into the privacy of the individual being tested,⁵⁰ thereby using a proportionality test in assessing the suitability of using genetic tests.⁵¹ As a basic principle, the legislation recognises the importance of employment and insurance in acting as a gateway to accessing further social goods and services.

2.2.6 Ireland

In Ireland this area is regulated primarily under the Disability Act 2005 and the Data Protection Acts 1998 and 2003. Part 4 of the Disability Act 2005 regulates genetic testing in a number of third party contexts, including insurance, employment and the mortgaging of property, by prohibiting the processing of genetic data in these contexts.⁵² In adopting a privacy approach, section 42 provides that the informed consent of the data subject is required in respect of the processing of genetic data.⁵³ It provides that the processing of genetic data is prohibited in relation to a policy of assurance, a policy of health insurance or health related product, an occupational pension, a retirement annuity contract or any other pension arrangement, unless the consent of the person has been obtained in accordance with the legislation.⁵⁴ The legislation therefore has a broad scope of application.

In the employment context, section 42 makes the processing of genetic data for employment an offence under the Data Protection Acts 1998 and 2003.⁵⁵ Under the data protection legislation, and specifically by virtue of the Data Protection

⁴⁹ Els Geelen *et al*, 'Unravelling fears of genetic discrimination: an exploratory study of Dutch HCM families in an era of genetic non- discrimination acts' (2012) 20 *European Journal of Human Genetics* 1018

⁵⁰ Law on Medical Examinations of 1997, Article 3

⁵¹ Trudo Lemmens, 'Selective Justice, Genetic Discrimination, and Insurance: Should we Single Out our Genes in Our Laws?' (2000) 45 *McGill Law Journal* 347, 360

⁵² Disability Act 2005

⁵³ *Ibid*, s 42(3)

⁵⁴ *Ibid*, s 42(2)

⁵⁵ *Ibid*, s 42(2)(a)

(Processing of Genetic Data) Regulations 2007⁵⁶ the processing of genetic data in relation to the employment of a person is now a prescribed activity for the purposes of section 12A of the Data Protection Acts 1988 to 2003.⁵⁷ Irish legislation offers relatively strong protections particularly in the employment context. There is an evident data protection approach taken in this part of the legislation.

2.3 Soft law approach: the UK

Soft law approaches are also identified in the EU. The moratorium adopted in the UK is particularly significant.

The UK's approach to genetic testing in the insurance context shows illustrates the operation of a moratorium. In consideration of the National Health Service, which is universally recognised as an effective public health care system, life insurance is the primary area of concern. The agreement between the Association of British Insurers and the Department of Health, known as the Concordat and Moratorium on Genetics and Insurance (first established in 2001) provides that individuals are not under an obligation to disclose results of genetic tests to insurers (up to a certain amount of insurance).⁵⁸ The moratorium has now been renewed further to 2017 and the next review will take place in 2014. It provides an exception in respect of life insurance policies in excess of £500,000 and only for government approved genetic tests.⁵⁹

The UK moratorium is unusual as it has been in place for many years. Generally a moratorium is introduced as a temporary solution until such time as a more permanent solution can be established. The fact that this has been a longer-term solution is perhaps illustrative of the success of this moratorium. Indeed the UK's moratorium has been described as an innovative and "*credible solution*" to the problem.⁶⁰

On examining the UK position in this area, it is necessary to have regard to recent legislative developments. In 2012 the Consumer Insurance (Disclosure and Representations) Act was introduced in the UK, which signals a new direction for insurance law in the UK, and in particular, the disclosure obligations on the insurance applicant.⁶¹ Pursuant to the legislation, individuals who misunderstand or misinterpret insurance questions or who misunderstand the

⁵⁶ Data Protection (Processing of Genetic Data) Regulations 2007 (S.I. No. 687 of 2007)

⁵⁷ See www.dataprotection.ie (accessed February 25, 2013)

⁵⁸ For further details see: <https://www.gov.uk/government/publications/agreement-extended-on-predictive-genetic-tests-and-insurance> (accessed 25 July 2013)

⁵⁹ Margaret Otlowski *et al*, 'Genetic Discrimination: International Perspectives' (2012) 13 Annual Review of Human Genetics 433, 446

⁶⁰ Yann Joly *et al*, 'Genetic Information and Life Insurance: A "Real" Risk?' (2003) 11 European Journal of Human Genetics 561, 563

⁶¹ Consumer Insurance (Disclosure and Representations) Act 2012

importance of their family history would not be severely punished for this non-disclosure, as was the position previously.⁶² The legislation will not have an effect on the existence of the moratorium, which will continue even in light of the legislative changes.⁶³ The legislation does however signal fundamental changes in insurance industry practices and principles.

2.4 Evaluation of the EU Member State responses – A patchwork of protection

Having highlighted the divergence in the modes of regulation and the different levels of protection in this area, through the analysis of some Member State laws, this section will draw some conclusions and consider the consequences that this variety import.

These diverging laws give rise to a patchwork of protection in this area. The most radical of these approaches is the prohibitive legislation which some states have enacted, specifically tailored to the issue of genetic testing. These laws offer relatively strong protections, primarily in the areas of employment and insurance. On the other hand, Ireland has incorporated protection under disability discrimination legislation, while the UK has adopted a voluntary moratorium. In addition, it is also apparent that some states take a non- discrimination approach, while others take a data protection approach.

Differences can also be identified in terms of the choice of regulatory model, and the choice of whether to enact a stand-alone law or include protections under other laws. In addition, there are differences as regards the material scope of the different laws, as well as differences in the definitions.⁶⁴ These stark differences and the resulting patchwork of protections created would seem to indicate that a uniform regulatory framework at EU level would be preferable, with a view to setting minimum standards in this area.

From a comparative perspective, it is also noted that the patchwork of protection at state level in the US was cited as a reason prompting the need for comprehensive federal level legislation to address the problems arising in this area.⁶⁵ In addressing the comparison between the US and the EU in this regard, the argument has been made that the US is a “*closer union*” than the EU and has a more harmonised and closely connected historical background.⁶⁶ In a well-connected union, there is a clear motivation to introduce federal level regulation to address the patchwork of legislation. In this context, it has been noted that the

⁶² Ruth Stirton, ‘Insurance, Genetic Information and the Future of Industry Self- Regulation in the UK’ (2012) 4 Law, Innovation and Technology 2 212, 236

⁶³ Ibid at 236

⁶⁴ Orsolya Varga *et al*, ‘Definitions of genetic testing in European legal documents’ (2012) 3 Journal of Community Genetics 125

⁶⁵ GINA, s 2(5)

⁶⁶ Janneke Gerards, ‘Regulation of Genetic Information in the United States’ in J.H. Gerards et al (eds) *Genetic Discrimination and Genetic Privacy in a Comparative Perspective* (Intersentia 2005) 113

EU does not have the same close connection and unity as the US in certain areas, such as employment and insurance.⁶⁷ However, it is noted that this is changing and the EU is moving closer towards unity. Increasingly borders are being eroded and employers, insurers (and other social and economic actors) are frequently conducting inter-state business. This is an added incentive to seriously consider the case for a uniform regulatory response to the issue of genetic information.

This patchwork of protection has a number of consequences. Firstly, it may potentially inhibit the effective operation of the internal market to a certain degree. For example, the internal market in the insurance industry may be adversely affected by the patchwork of laws in the Member States. It may create burdensome administrative, legal and other costs for insurance companies operating in numerous Member States. The employment market may also be negatively affected by this patchwork of laws, particularly for employers who operate across various Member States, as well as for employees who cross borders. As regards this internal market argument, it is acknowledged that the patchwork of laws situation really only becomes a problem and seriously impacts upon the internal market when it affects large- scale business and commerce, particularly when operating across borders. However, it is prudent to anticipate such problems at an early stage.

Further, this divergence in laws and the radical differences in the levels of protections may jeopardise the movement of people within the EU. One of the core objectives of the EU is to ensure the unfettered movement of people throughout the Member States.⁶⁸ Linked to this, it is also acknowledged that the advancement of employment is a core objective of the EU.⁶⁹ Divergences as regards the accessibility of the employment market may hinder the movement of individuals, and particularly individuals with disabilities (and putative disabilities) in the EU. Similarly, divergences as regards the insurance market may present barriers to accessing a range of further social goods and services, particularly for persons with disabilities (and persons with putative disabilities). The free movement of goods, persons and services within the EU is key to ensuring the functioning of the internal market. It is also noted that the patchwork of laws creating barriers to the internal market also negatively impacts upon the economic participation of persons with putative disabilities. For example, barriers to accessing and maintaining employment has a key impact on the economic activity of such individuals, as do barriers to accessing insurance. Society may lose out on the economic contribution of persons with putative disabilities.

As illustrated in chapter 3, the existence of barriers in accessing employment and insurance also impacts economic activity in other facets of life, such as social

⁶⁷ Ibid at 113

⁶⁸ Article 3(2) TEU

⁶⁹ Aart Hendriks, '(Pre-) employment Medical Examinations and the Law, with Particular Reference to the European Union' (1994) 1 European Journal of Health Law 229, 232

and cultural life. In this regard, it is recognised that the patchwork of laws distorts the achievement of human rights in the EU. In light of the fact that the EU is moving from being primarily focused on the economic objectives of the union, towards embracing both the core economic priorities, as well as human rights principles, there is an added incentive to include human rights within this discussion. Pursuant to this dual objective, the EU is obliged to remove the various economic, social and other barriers which prevent the operation of the internal market and prevent the fulfillment of human rights of all individuals.

3. Public confidence in science and technology

In addition to building a case for EU level action in this area based on the current patchwork of laws, a key argument made in support of such action is to enhance public confidence in advancing science and technology. The rationale of this argument is that preemptive legislation is needed in the EU to enhance and maintain public confidence in advancing genetic science and technology. Public trust relates to both trust in the advantages of such technologies and confidence that there are adequate legal protections in place. This submission is in line with the importance of science, technology and innovation in the EU and the importance of a knowledge economy, as emphasised in *Europe 2020*.⁷⁰ From a research and development perspective, these objectives are also reflected in *Horizon 2020* and the current research framework in the EU.⁷¹ It is further pointed out that “*technological advance has been regarded as a prerequisite for improving social and economic progress since modern times.*”⁷²

It is submitted that the goal of enhancing public confidence and progressing science also serves the objective of facilitating and sustaining a viable economy. It acknowledges the value of a knowledge economy and acknowledges the impact which science and technology has on the economy. Particularly in times of economic recession, there is additional importance attached to this objective in the EU. It is recognised that there is a connection between public confidence, scientific progression and subsequent economic gains. This connection needs to be acknowledged when considering an EU level response in this area.

As has been illustrated in chapter 2, science and technology are advancing rapidly. However, there is also a corresponding potential for misuse. The fear of the misuse of one's genetic information, results in some individuals being

⁷⁰ Communication from the Commission, 'Europe 2020 – A Strategy for smart, sustainable and inclusive growth' COM (2010) 2020 final. See also Brian Wynne et al, 'Taking European Knowledge Society Seriously' (European Commission 2007)

⁷¹ Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions: Horizon 2020 – The Framework Programme for Research and Innovation COM (2011) 808 final

⁷² Maria Eduarda Gonclaves and Maria Ines Gameiro, 'Does the Centrality of Values in the Lisbon Treaty Promise More Than it Can Actually Offer: EU Biometrics Policy as a case study' (2013) (first published online 4 June 2013) 1, 2

reluctant to take advantage of advancing genetic technology.⁷³ This reluctance to engage in genetic testing may also have the effect of stifling genetic science and advancing technology. Scientists may therefore experience difficulty in collecting research data as people become unwilling to submit to testing and engage in clinical trials. This may have further implications for public health, as potentially beneficial genetic discoveries and technological innovations become hampered. Indeed this concern has been recognised in a major EU policy document, '*Taking European Knowledge Society Seriously*.'⁷⁴ This document acknowledges the general societal fear of scientific and technological developments. This fear needs to be addressed in order for society to fully reap the benefits of such developments.⁷⁵

As discussed in chapter 7, one of the objectives behind GINA was to enhance public confidence in genetic science and technology. Indeed, prior to GINA, it was opined, "*Unless Americans are convinced that the information will not be used against them, the era of personalized medicine will never come to pass.*"⁷⁶ The legislation was "*necessary to fully protect the public from discrimination and allay their concerns about the potential for discrimination, thereby allowing individuals to take advantage of genetic testing, technologies, research, and new therapies.*"⁷⁷ In this regard, reference is made to a report on the economic value and impact of the Human Genome Project. It has been found that genetic developments and the genomics industry is facilitating further scientific innovation and commercial results in a range of areas.⁷⁸ It is arguable that the US legislative endeavours have in some way contributed to this success and public confidence. Although in this regard, it is acknowledged that there are still challenges to be faced in the US as regards public confidence, as noted in chapter 7.

In line with the EU's goals in this area, it is submitted that an EU level response would contribute towards promoting public confidence in the field of genetic science and technology, as well as facilitating the EU's research and development objectives. Having discussed the implications of the patchwork of current protections and the need to maintain public confidence in science and technology, the following section will highlight the consequent imperative for a uniform EU level response.

⁷³ Louise Slaughter, 'The Genetic Information NonDiscrimination Act: Why your personal genetics are still vulnerable to discrimination' (2008) 88 *The Surgical Clinics of North America* 4 723, 737

⁷⁴ Brian Wynne *et al*, '*Taking European Knowledge Society Seriously*' (European Commission 2007)

⁷⁵ *Ibid*

⁷⁶ Francis Collins, 'The Threat of Genetic Discrimination to the Promise of Personalized Medicine', Testimony before the Subcommittee on Health Committee on Energy and Commerce, United States House of Representatives (9 March 2007)

⁷⁷ GINA, s 2(5)

⁷⁸ Battelle Memorial Institute, Report: 'The Impact of Genomics on the US Economy' (June 2013), available at: <http://www.battelle.org/docs/health-and-pharmaceutical/the-impact-of-genomics-on-the-u-s-economy-june-11-final.pdf?sfvrsn=0> (accessed 14 August 2013). See also previous report: 'Economic Impact of the Human Genome Project' (May 2011)

4. Towards an EU level response

The proposal for an EU level response is also made in recognition of the fundamental human rights at stake, the competing interests arising and the need to regulate the use of genetic information. Building upon the analysis provided throughout, and on consideration of the existing patchwork of protection highlighted, as well as the need to maintain public confidence in science and technology, this thesis proposes consideration of an EU level response to address the discriminatory use of genetic information. The following sections will further highlight the case for an EU level response and will explore the legal basis for such action.

As discussed in chapter 5, this thesis is in favour of a genetic specific approach towards regulation of this area. In this regard, it proposes consideration of a genetic specific approach at EU level. As highlighted in chapter 5, such an approach reflects, amongst other things, the unique, personal and valuable nature of genetic information which provide compelling reasons behind the adoption of special protections to regulate genetic information. Further, such an approach is proposed in consideration of existing incomplete protections available at EU level. In addition, from the perspective of ensuring and maintaining public confidence in advancing genetic science and technology, it is proposed that a genetic specific approach at EU level carries more weight and expressive value.

In chapter 5, the analysis of the regulatory models indicated that both the non-discrimination and privacy (data protection) models offer an appropriate framework from which to address this area. This provoked consideration of the possibility of a two- pronged approach to address this area based on the non-discrimination and data protection frameworks. However, in light of the fact that there is currently a major reform process underway in the EU in the area of data protection law (see chapter 9), it is argued that the most appropriate and practical framework to consider here is a stand- alone non- discrimination one. As highlighted in chapter 9, the current data protection reform process is a welcomed development in this area. As regards the regulation of genetic information, this provides the foundation step to consider a complementary regime, such as non- discrimination. Such a complementary approach would ensure that access to genetic information is controlled and the discriminatory use of genetic information is prohibited.

In light of the EU constitutional framework, it is therefore necessary to address whether the EU has competence to legislate on the discriminatory use of genetic information. In this regard, this thesis proposes consideration of a dual legal basis to ground the EU's competence to act. It proposes consideration of Article 19 Treaty on the Functioning of the European Union ("TFEU"), which provides the power to address discrimination, on a number of grounds including disability,

as well as Article 114 TFEU which provides the power to act to ensure the functioning of the internal market. This will require an examination of the applicable EU Treaty articles. It will also consider the value of the CFREU and the CRPD as an impetus to act in this area. Although these instruments do not extend the competences of the EU, they are nevertheless important in supporting the proposal for EU action in this area.

4.1 Competences of the EU and legal basis for action

Before exploring the specific legal bases for action, this section will briefly highlight the general EU competence principles. Since the EU was established, key competences have moved from Member States to the EU, thereby reducing and limiting the individual competences of the Member States, and increasing the competences of the EU.⁷⁹

The majority of competences are shared between the Member States and the EU and must be exercised in compliance with the principles of subsidiarity and proportionality.⁸⁰ Pursuant to the principle of subsidiarity, in areas of shared competence, EU action in that area is only permissible in circumstances where the MS cannot achieve the goals of the Treaties and the EU can carry out these goals in a more efficient and effective manner.⁸¹ Pursuant to the proportionality principle any such legislative action by the EU “*shall not exceed what is necessary to achieve the objectives of the Treaties.*”⁸² It is important to have general regard to these principles when considering legislative endeavours by the EU, and when considering whether certain legislative actions come within the scope of the EU’s powers to act.

Pursuant to Article 4 TFEU, the areas of economic and social cohesion, as well as the regulation of the internal market, are all areas of shared competence.⁸³ Accordingly, the principles of subsidiarity and proportionality need to be satisfied. These principles will be further addressed below. The following sections will explore the scope of the non- discrimination and internal market provisions of the treaties.

4.2 Non- Discrimination in the EU

On examining the legal basis for non- discrimination action in the EU, it is noted that the principle of non- discrimination and principles of equality are clearly provided and emphasised in the Treaties. The EU has a clear competence in the

⁷⁹ Alina Kaczorowska, *European Union Law* (2nd edn, Routledge 2011) 80

⁸⁰ Article 5 TEU

⁸¹ Alina Kaczorowska, *European Union Law* (2nd edn, Routledge 2011) 87

⁸² Article 5(4) TEU

⁸³ Article 4 TFEU

field of non- discrimination on a number of grounds. There are several articles which mention the principle of non- discrimination and equality.

Firstly, the core values, and the aims and objectives of the EU are set out Article 2 and Article 3 of the Treaty on the European Union (“TEU”) and there is an evident focus on human rights. Article 2 refers to a number of societal values, including non- discrimination, upon which the EU is founded.⁸⁴ Article 3 sets out the aims and objectives of the EU, which include, *inter alia*, non- discrimination and combating social exclusion, as well as promoting social justice and protection.⁸⁵ The inclusion of these provisions in the TEU confirms the importance of upholding fundamental human rights, and addressing discrimination as core objectives.

The human rights framework of the EU is incorporated into the TEU. In light of the human rights approach taken in this thesis and in view of the reference to the CFREU and its impact on the EU human rights framework, Article 6 TEU is noted. As highlighted in chapter 9, this provision recognises the principles set out in the CFREU and gives the Charter the same value as the Treaties. The EU is now obliged to comply with the Charter in all its activities. The importance of the CFREU and the relevant non- discrimination provisions therein, most notably Article 21(1) have been discussed in chapter 9. Although it is noted that the CFREU can inform EU level legislation, it is emphasised that Article 6 does not change or enhance the competences of the EU.

On setting out the nature and scope of the EU’s competences, it is noted that the TFEU focuses not only on the economic objectives of the union, but also on the aim of ensuring social inclusion, equality and other fundamental rights. Article 10 TFEU contains a non- discrimination clause. It states, “*in defining and implementing its policies and activities, the European Union shall aim to combat discrimination based on sex, racial or ethnic origin, religion or belief, disability, age or sexual orientation.*”⁸⁶ Pursuant to this provision, it is therefore intended that addressing discrimination on the grounds of, *inter alia*, disability is a core priority of the EU in implementing policies.

Article 19 (formerly Article 13) provides the power for the EU to: “... *take appropriate action to combat discrimination based on sex, racial or ethnic origin, religion or belief, disability, age or sexual orientation...*”⁸⁷ The objective of Article 19 is “*to protect the dignity and autonomy of persons belonging to those suspect classifications.*”⁸⁸ Article 19 was therefore intended to fulfill a range of core human rights. Article 19 was a new departure in the area of disability

⁸⁴ Article 2 TEU

⁸⁵ Article 3 TEU

⁸⁶ Article 10 TFEU

⁸⁷ Article 19 TFEU

⁸⁸ Opinion of Advocate General Maduro, Case C- 303/06 *Coleman v Attridge Law & Steve Law* [2008] ECR I-5603, p 10

discrimination law in providing for the power to act and legislate to address discrimination on the grounds of 'disability.' This was an important development in EU human rights law and it enhanced the power of the EU to act in the area of disability discrimination. This article has great significance in terms of ensuring inclusion and equality of opportunity for all EU citizens. Indeed, the EU has relied upon Article 19 in enacting a number of EU Directives.

4.2.1 'Article 13 Directives'

By way of brief illustration of the potential of Article 19, this section will highlight its application. The adoption of the 'Article 13 Directives' (as discussed in chapter 9) highlight the EU's commitment to principles of non- discrimination, and ensuring the protection of fundamental human rights.⁸⁹ These legislative endeavours confirm the EU's competence to legislate in the area of discrimination, on the basis of Article 19. These actions also confirm the EU's competence to legislate in the particular area of disability discrimination. Particular reference is also made to the Race Directive, which segregates the issue of race in a stand- alone directive. It is a useful comparable model to base consideration of a genetic information non- discrimination directive.

On evaluation of Article 19, the emphasis on human rights is clear and it is a confirmation of the EU's commitment to incorporating a strong human rights framework into the EU's constitutional framework. In this regard, it has been asserted, "*as the Community moves into the next millennium, it will be of the greatest importance, in creating a Union truly founded on human rights.*"⁹⁰ Within this framework, the inclusion of strong non- discrimination powers provide an effective means of achieving and fulfilling such rights, as reflected in these directives. The principle of non- discrimination is used in these circumstances to ensure that all individuals in the EU enjoy equality of opportunity and freedom from discrimination, including persons with disabilities.

4.2.2 Article 19 and genetic information

It is submitted that Article 19 TFEU offers a viable basis for the introduction of non- discrimination legislation on the grounds of genetic information. Although Article 19 itself does not mention 'genetic information' as a discrimination ground, it could be a suitable legal base through an expansive interpretation of the ground of disability (to include imputed disability, future disability or genetic predisposition to disability). Ferri has observed the importance of this provision in the context of clarifying the EU's competence in the field of disability

⁸⁹ See chapter 9

⁹⁰ Robin Allen QC 'Article 13, evolution and current contexts', in Helen Meenan (ed) *Equality Law in an Enlarged European Union – Understanding the Article 13 Directives* (Cambridge University Press 2007) 58

discrimination.⁹¹ An expansive interpretation of Article 19 could arguably encompass the inclusion of discrimination on the basis of imputed disability, future disability and therefore discrimination on the basis of genetic predisposition to disability.

This interpretation takes a social model approach to the definition of disability and one that is in accordance with the CRPD. We can look to recent case law of the European courts as reference for this modern definition of disability. The joined case of *Ring and Werge*⁹² interpreted the definition of disability in light of the CRPD, as did the recent case of *Commission v Italy*.⁹³ Further, the case of *Coleman v Attridge*, (also discussed in the chapter 9) expands the interpretation of the disability ground, to cover not just those who themselves have a disability, but those relatives who may find themselves discriminated against 'on the basis of disability'.⁹⁴

In these cases, the court therefore acknowledged the various external barriers that operate to disable an individual and inhibit their full and effective participation in society. In this regard, there is also a focus on the notion of discrimination 'on the basis of disability,' which places the emphasis on the actions and intention of the discriminator, and takes the focus away from the individual. It is submitted that this social construction of disability incorporates, not only the concept of future disability but also the concept of imputed disability, and genetic predisposition to disability.

In addition, as discussed in chapter 6, the conclusion of the CRPD by the EU provides an added dimension to the debate of regulating genetic information. Although not creating any strict legal obligations to act in these circumstances, it certainly acts as an impetus to action in this area, and acts as an international standard from which to view the issue of disability discrimination. Further, the conclusion of the CRPD by the EU offers the opportunity to take a more proactive approach in terms of bringing the EU in line with the obligations under the CRPD.

Such an interpretation of Article 19 also resonates with the CFREU. The specific reference to 'genetic features' in Article 21(1), and the additional value placed on the Charter by the Lisbon Treaty, strengthens the argument that the EU not only has the power to legislate in the area of genetic information and discrimination (by virtue of Article 19) but also has obligations to comply with the principles of the Charter in enacting legislation where required. It has also been observed that

⁹¹ Delia Ferri, 'Legislating at EU level: Challenges and Possibilities' (Genetic Discrimination - Transatlantic Perspectives on the Case for a European Level Legal Response, conference, Galway, Ireland, 19 November 2011)

⁹² Joined cases C- 335/11 and C- 337/11 *HK Danmark, acting on behalf of Jette Ring v Dansk almennyttigt Boligselskab* and *HK Danmark, acting on behalf of Lone Skouboe Werge v Dansk Arbejdsgiverforening, acting on behalf of Pro Display A/S, in liquidation* (11 April 2013)

⁹³ Case C-312/11 *European Commission v Italy* [2013] (4 July 2013)

⁹⁴ Case C-13/06 *Coleman v Attridge Law*, [2008] ECR I 5603. The insights of Aart Hendriks in critiquing this case are acknowledged – see chapter 9 for further discussion.

such a wide interpretation would also be in line with other fundamental rights provided for in the Charter, for example, the right to dignity and integrity of the person.⁹⁵ In addition, pursuant to the analysis in chapter 9, the CRFEU can perhaps prompt the EU to reflect upon the case for EU level regulation of genetic information. This interpretation is also in accordance with the jurisprudence of the ECHR, which is beginning to recognise the ground of genetic predisposition to disability and protection of genetic information.⁹⁶

It is acknowledged that Gerards and Janssen also tentatively considered Article 19 (or Article 13 as it was known) as a potential legal basis for regulating genetic information at EU level.⁹⁷ However, it is noted that these submissions were made almost ten years ago and EU non- discrimination has evolved significantly since then, particularly in the area of disability, as illustrated above. Gerards and Janssen also noted challenges as regards the principle of subsidiarity, with particular reference to the potential harm to national traditions.⁹⁸

On balance, it is submitted that Article 19 would provide a suitable basis for enacting legislation addressing discrimination on the grounds of genetic information or genetic predisposition to disability, based on the above analysis. There is a clear, legally sound basis for considering such legislation.

4.3 The internal market

In conjunction with the need to tackle discrimination, it is also submitted that another core EU objective merits consideration on looking towards EU regulation of genetic information. This objective is the achievement and facilitation of the internal market, which is provided for in Article 114 TFEU.⁹⁹ On highlighting the merits of an internal market approach, reference is made to *Europe 2020*. In this document, the interdependence and the close links of the EU Member States is emphasised.¹⁰⁰ With the intention of fulfilling the economic objectives of the EU, Article 114 permits the enactment of legislation to facilitate the functioning of the internal market in the EU.

⁹⁵ Delia Ferri, 'Legislating at EU level: Challenges and Possibilities' (Genetic Discrimination - Transatlantic Perspectives on the Case for a European Level Legal Response conference, Galway, Ireland, 19 November 2011)

⁹⁶ *S. and Marper v. the United Kingdom* (No. 30562/04) [2008] ECHR 1581 4 December 2008, *G.N. and others v Italy* (No. 43134/05) [2009] ECHR 1 December 2009

⁹⁷ Janneke H. Gerards and Heleen L. Janssen, 'Regulation of Genetic and Other Health Information in a Comparative Perspective' (2006) 13 *European Journal of Health Law* 339, 372- 374

⁹⁸ *Ibid*

⁹⁹ Article 114 TFEU

¹⁰⁰ Communication from the Commission, 'Europe 2020 – A Strategy for smart, sustainable and inclusive growth' COM (2010) 2020 final

Article 114 previously acted as the legal basis for various pieces of legislation, for example Directive 95/16/EC,¹⁰¹ and Directive 2001/85/EC.¹⁰² It has been noted that these legislative endeavours acknowledge “*the disability dimension to securing the internal market*.”¹⁰³ The rationale and premise underlying this legal basis is to facilitate the operation of the internal market and to ensure the elimination of any potential obstacles to the functioning of the internal market. It effectively aims at harmonising the laws of the EU in particular areas. The EU uses the internal market approach to legislate to effectively eliminate a fragmented approach to legislation by the Member States.

In the context of regulating genetic information and using the internal market as a legal basis, it is observed that the current patchwork of legal protections potentially inhibits the proper functioning of the internal market. As highlighted, there is an evident divergence in the legislative and policy approaches adopted, with varying degrees of protection available against the misuse of genetic information. The diverging approaches towards regulation of genetic information may potentially act as an obstacle to the freedom of movement of workers and persons within the EU, as noted above. It may also act as an obstacle to the freedom of movement of services within the EU, for example, in respect of the insurance industry. These collective obstacles may further inhibit the functioning of the internal market and the economic activity of individuals in the EU.

Similarly, it is also submitted that obstacles in accessing and maintaining employment and insurance hinder the economic and social activity of persons with disabilities, specifically, persons with putative disabilities, which includes a large portion of the EU population. Further, in light of the ageing population and the increasing advances in genetic science and technology, more individuals can be categorised as having putative disabilities. Therefore ensuring the effective operation of the internal market in this area may enhance the participation and inclusion of such persons in EU society and the economy.

It is observed that in the US, the legal basis for enacting the Genetic Information Nondiscrimination Act 2008 was the commerce clause (as noted in chapter 7).¹⁰⁴ It was acknowledged that the existing patchwork of laws that was apparent throughout the US inhibited the operation of interstate commerce, therefore prompting the need for a federal level law, in the form of GINA. Indeed, such a patchwork is referred to in the legislation.¹⁰⁵

¹⁰¹ Directive 95/16/EC on the approximation of the laws of the Member States relating to lifts [1995] OJ L213/1 as amended

¹⁰² Directive 2001/85/EC relating to special provisions for vehicles used for the carriage of passengers comprising more than eight seats in addition to the driver’s seat, and amending Directives 70/156/EEC and 97/27/EC, [2002] O.J. L.43/1

¹⁰³ Lisa Waddington, ‘A New Era in Human Rights Protection in the European Community: The Implications the United Nations Convention on the Rights of Persons with Disabilities for the European Community’ (2007) 4 Maastricht Faculty of Law Working Paper, 11

¹⁰⁴ Constitution of the United States, Article 1, section 8, clause 3

¹⁰⁵ GINA, s 2(5)

The recent proposals for the EU Accessibility Act are also based upon the internal market argument and the EU's obligations to achieve and maintain this. This legislative proposal aims to improve the accessibility of goods and services in the internal market, and focuses on the removal of barriers for persons with disabilities.¹⁰⁶ Further, this legislative proposal submits that the barriers to accessing goods and services in the EU, as a result of a lack of accessibility, inhibits the economic, social and political participation in society for persons with disabilities.¹⁰⁷ These obstacles to such economic, social and political integration therefore operate to further restrict and inhibit the internal market within the EU.

This thesis submits that Article 114 is a suitable legal basis upon which to regulate genetic information in the EU, with particular reference to the current patchwork of laws which exists in the EU and the adverse impact upon the internal market, as well as the focus on the economic inclusion and activity of persons with putative disabilities.

Consideration of the legal basis for action at EU level provokes the question as to whether it might be preferable to recommend a double legal basis – Article 19 on non- discrimination, together with Article 114 on the internal market. The following section will briefly look at the merits of such an approach.

4.5 Combining the economic and human rights objectives of the EU

When the EU (European Community) was first established, the core objectives focused on the economic activities of the union, and securing the effective operation of the European market. However, it is becoming increasingly apparent that there is a move towards greater cohesion between the economic objectives of the EU and the objective to fulfill human rights.¹⁰⁸ For example, it has been stated that *"it will be necessary for the EU... to move beyond an instrumental, ad hoc, market- led mentality towards a mature conception of fundamental rights as goods in themselves."*¹⁰⁹

This submission is supported by the evident move towards recognising and adopting human rights instruments in the EU. For example, the conclusion of the CRPD by the EU and the enhanced value of the CFREU, which further promotes the inclusion of human rights within the EU legal framework and political agenda.

¹⁰⁶ See EU Accessibility Act roadmap:

http://ec.europa.eu/governance/impact/planned_ia/docs/2012_just_025_european_accessibility_act_en.pdf (accessed 22 August 2013)

¹⁰⁷ Ibid

¹⁰⁸ Maria Eduarda Gonclaves and Maria Ines Gameiro, 'Does the Centrality of Values in the Lisbon Treaty Promise More Than it Can Actually Offer: EU Biometrics Policy as a case study' (2013) (first published online 4 June 2013) 1

¹⁰⁹ Sionaidh Douglas- Scott, 'The European Union and Human Rights after the Treaty of Lisbon' (2011) 11 Human Rights Law Review 4 645, 681

Further, the future accession of the EU to the ECHR is an added indication of the incorporation of human rights into the EU legal system.¹¹⁰

In the context of considering a dual legal basis, reference is made to Council Decision on the conclusion of the CRPD by the EU.¹¹¹ The legal basis for this instrument was both Article 13 (Article 19) and Article 95 (Article 114). This is a further illustration of the increasing cohesion between the economic objectives and the human rights objectives of the EU. It also illustrates the recognition of combining the regulation of disability with facilitating the operation of the internal market, as well as emphasising the importance of the economic contribution of persons with disabilities.

It is submitted that this evolution in the EU political and legal framework would support the adoption of a dual legal basis, on the grounds of Article 19 and Article 114, to promote the economic aims of the union, enhance the operation of the internal market, and to address the issue of discrimination on the grounds of genetic information, (or genetic predisposition to disability). Both goals can be achieved through the enactment of legislation to regulate genetic information in the employment and insurance contexts.

This thesis also acknowledges the EU's competence to act in the fields of employment and private insurance. The EU has clear competence to act in the field of employment, as is provided in Article 5 TFEU. In highlighting the EU's competence to act in this area, it is noted that there have been numerous EU directives introduced, including the Employment Equality Directive, as discussed. The field of private insurance traditionally came within the responsibility of the Member States, pursuant to the principle of subsidiarity.¹¹² However it is noted that it is gradually coming within the scope of the EU's competence and objectives. In terms of highlighting the EU's competence to act in this area, it is noted that the EU has introduced a series of Directives over the past number of years, with the objective of formulating an internal market in insurance services.¹¹³ It has been observed that pursuant to the principle of the free

¹¹⁰ Article 6 (2) TEU

¹¹¹ Council Decision of 26 November 2009 concerning the conclusion, by the European Community of the United Nations Convention on the Rights of Persons with Disabilities (2010/48/EC) [2010] OJ L

¹¹² See generally, Sarah Thomson and Elias Mossialos, 'Regulating Private Health Insurance in the European Union: the implications of single market legislation and competition policy' (November 2006, London School of Economics and Political Science) Working Paper No. 4/2006. See also Sarah Thomson and Elias Mossialos, 'EU law and regulation of private Health insurance' (2007) 2 Health Economics, Policy and Law 117

¹¹³ Sarah Thomson and Elias Mossialos, 'Private health insurance and the internal market' in Elias Mossialos et al (eds) *Health Systems Governance in Europe* (Cambridge University Press 2010) 419, at 426. See First Council Directive 73/239/EEC on the coordination of laws, regulations and administrative provisions relating to the taking up and pursuit of the business of direct insurance other than life insurance OJ 1973 No. L228/3; Second Council Directive 88/357/EEC on the coordination of laws, regulations and administrative provisions relating to direct insurance other than life insurance and laying down provisions to facilitate the effective exercise the freedom to provide services and amending Directive 73/229/EEC, OJ 1988 No. L171/1; Council Directive

movement of services,¹¹⁴ the objective and intention of the internal market in insurance services is to promote competition and improve consumer choice.¹¹⁵ EU competence to act in this area derives from the fact that insurance is deemed to be an economic activity.¹¹⁶ In illustrating an EU framework for regulating private health insurance, particular reference is made to the Third Non- Life Insurance Directive which was introduced in 1992 and which highlights EU's competence in this area.¹¹⁷

5. Evidence of genetic discrimination

On building a case for EU level action, it is also necessary to refer to the documented evidence of genetic discrimination. This section will highlight some of the case law and other evidence indicating that genetic discrimination and fear of genetic discrimination is a problem. It acknowledges that, currently, there is very little evidence of genetic discrimination and breach of genetic privacy taking place in the EU, either in terms of case law or empirical evidence. The argument could therefore be made that there is not yet a problem of misuse of genetic information taking place.

As regards case law, reference is made to the documented cases in the US, as discussed in chapter 7.¹¹⁸ In the EU, there have not been as many cases on the direct issue of genetic discrimination. In 2006 a case was reported from Germany which concerned a female teacher who was discriminated against in her employment as a result of a family history of Huntington's disease.¹¹⁹ On being refused a permanent job on the basis of her family history, the woman disputed the decision to the German courts, and won her case.¹²⁰ Although this case does not indicate a prevalence of genetic discrimination or discrimination on the grounds of family medical history, it does serve as a useful example of the potential for such invidious discrimination in Europe.

92/49/EEC on the coordination of laws, regulations and administrative provisions relating to direct insurance other than life insurance OJ 1992 L.228/23

¹¹⁴ Articles 56- 57 TFEU

¹¹⁵ Ibid at 426

¹¹⁶ Ibid at 426

¹¹⁷ Council Directive 92/49/EEC on the coordination of laws, regulations and administrative provisions relating to direct insurance other than life insurance OJ 1992 L.228/23. See Sarah Thomson and Elias Mossialos, 'EU law and regulation of private Health insurance' (2007) 2 Health Economics, Policy and Law 117

¹¹⁸ For example, *Norman- Bloodsaw v Lawrence Berkeley Laboratory*, 135 F.3d 1260, 1265 (9th Cir. 1998) at 1271-72. See also Press Release, EEOC, 'EEOC Settles ADA Suit Against BSNF for Genetic Bias (18 April 2001) available at: <http://www.eeoc.gov/press/4-18-01.html> (accessed 26 July 2013). See also recent cases: *Equal Employment Equality Commission v Fabricut Inc.* Case No.: 13-cv-248-CVE-PJC (filed 7/05/2013), *Equal Employment Equality Commission v Founders Pavilion Inc.d/b/a Founders Pavilion* Case No. 6:13-cv-06250-CJS (filed 16/05/2013)

¹¹⁹ Dagmar Schmitz and Urban Wiesing, 'Just a Family Medical History? (2006) 332 British Medical Journal 297. See also Jane Burgermeister, 'Teacher was refused job because relatives have Huntington's disease' (2003) 327 British Medical Journal 827

¹²⁰ Ibid

In terms of evidence of genetic discrimination and evidence of fear of genetic discrimination, it is observed that there has been very little empirical evidence gathered, particularly in the EU. In 1992 Billings *et al* reported the first alleged case studies of genetic discrimination in the US.¹²¹ This study involved 29 people who had experienced adverse treatment allegedly on the basis of their genetic characteristics in the US and Canada. The study reported that genetic discrimination exists especially in the areas of health and life insurance.¹²² In 1996 Geller *et al* reported genetic discrimination incidences in a case study involving at-risk individuals and children in the US.¹²³ This study found that genetic discrimination occurred against asymptomatic individuals in a number of areas including in health and life insurance, adoption agencies, and in education.¹²⁴ In 1996 Lapham *et al* published a survey which found that a large number of respondents were refused health and life insurance, and were not hired or were dismissed from work on the grounds of having a genetic disorder.¹²⁵

The majority of the studies carried out were in the US. In Europe, there is very little evidence; particularly evidence which verifies misuse in employment, insurance or other fields. In addition, any studies that were carried out are likely to be somewhat redundant now, in light of technological advances in recent years and the greater accessibility to genetic information. In the life insurance context, Low *et al* evaluated experiences between individuals with a genetic condition and the general population and found that those with genetic conditions experienced more genetic discrimination.¹²⁶

In 2003 GeneWatch UK published a report, '*Genetic Testing in the Workplace*' which looked at the potential for misuse of genetic information in employment.¹²⁷ While it did not gather empirical evidence of misuse, it did investigate the use of genetic information and the issues that may arise. It found that genetic testing is not scientifically valid to use in employment. It also recommended that legislation needs to be introduced to prohibit employers (and insurers) from accessing or using genetic information.¹²⁸ More recently, a study from Germany, which

¹²¹ Paul Billings *et al*, 'Discrimination as a consequence of genetic testing' (1992) 50 American Journal of Human Genetics 476

¹²² Ibid

¹²³ Lisa Geller *et al*, 'Individual, family and societal dimension of genetic: A case study analysis (1996) 2 Science and Engineering Ethics 1 71

¹²⁴ Ibid

¹²⁵ Virginia E. Lapham *et al*, 'Genetic Discrimination: Perspectives of Consumers' (1996) 274 Science 621

¹²⁶ Lawrence Low *et al*, 'Genetic discrimination in life insurance: empirical evidence from a cross sectional survey of genetic support groups in the United Kingdom' (1998) 317 British Journal of Medicine 1632

¹²⁷ Report, 'Genetic Testing in the Workplace' (GeneWatch UK, June 2003). See also Susan Mayor, 'Report warns of danger of genetic discrimination in the workplace' (2003) 327 British Medical Journal 702

¹²⁸ Ibid

focused on Huntington's disease, reported several instances of genetic discrimination, as well as a fear of genetic discrimination.¹²⁹

It has been noted that the most of these reports, "*were limited to case studies or third- hand reports, and were typically based on the presence of disease in contrast to genetic predisposition.*"¹³⁰ Therefore, to a certain extent, the value is limited. Australia has offered the best example of empirical evidence in this area and in 2002 the first major national investigation of genetic discrimination was carried out, (see chapter 7).

In addition to documented evidence of genetic discrimination, there have also been studies indicating fears of genetic discrimination, in particular confirming a fear of misuse of genetic information by interested third parties. These studies have taken place mainly in the US and Canada. In 2005, a study involving hemochromatosis was carried out in the US and Canada, which showed that almost half of those surveyed were worried that genetic testing may result in difficulties in securing or maintaining insurance.¹³¹ This study also noted the potential that fear of discrimination may inhibit participation in research, thereby stifling science and technology. Another study indicating fear of discrimination involved those with Huntington's disease.¹³² It is noted that there is an absence of these studies in the EU indicating fear of genetic discrimination.

The analysis of the evidence of genetic discrimination prompts discussion of the potential challenges that might arise in proposing EU level action in this area.

6. Potential challenges to EU action

This section will highlight the potential challenges to proposing an EU level response to address the discriminatory of genetic information. This includes an examination of the restrictions on the competence of the EU, as well as the lack of firm evidence of genetic discrimination and breach of genetic privacy in the EU. This section will also address the principles of subsidiarity and proportionality.

6.1 Lack of evidence of misuse of genetic information

The argument could be made that there is not yet a problem of misuse of genetic information taking place. Indeed, this argument was made in the US by opponents of GINA who questioned the necessity of the legislation (as noted in

¹²⁹ Thomas Lemke, "A slap in the face." An exploratory story of genetic discrimination in Germany' (2009) 5 Genomics Law and Policy 2 22

¹³⁰ Margaret Otlowski *et al*, 'Genetic Discrimination: International Perspectives' (2012) 13 Annual Review of Genomics and Human Genetics 433, 438

¹³¹ Mark Hall *et al*, 'Concerns in a primary care population about genetic discrimination by insurers' (2005) 7 Genetics in Medicine 311

¹³² Yvonne Bombard *et al*, 'Beyond the patient: the broader impact of genetic discrimination among individuals at risk of Huntington disease' (2012) 159B American Journal of Medical Genetics 2 217

chapter 7). The above analysis points to a gap in the evidence of genetic discrimination in the EU, and a need for solid empirical evidence. This is in comparison to Australia, where a significant investigation took place, which gathered evidence of incidents of misuse of genetic information in a variety of scenarios. This study proved effective in informing the law reform process and the legislative endeavours that are taking place in Australia in this area. It is likely that in the event of proposing legislation at EU level, there will be a need to put forward a convincing argument that the use and potential misuse of genetic information is creating problems, causing harm and breaching fundamental human rights. In addition, from a scientific perspective, it may also be necessary to present evidence to show that the lack of comprehensive regulation at EU level operates to stifle scientific innovation and technological development. This may indicate a need to gather evidence of public attitudes in this area.

Nevertheless, even in the absence of empirical evidence of genetic discrimination in the EU, it is submitted that there is a need for a proactive, preemptive approach in this area, in light of the speed at which science and technology is advancing and the myriad of fundamental human rights at stake, as was the case in the US. Proponents of the legislation in the US saw this lack of evidence as an opportunity to address this area before it becomes a more widespread problem in the future. The evidentiary hurdle is recognised, but it is nevertheless submitted that there is merit in taking such a proactive approach. It is also noted that the existing evidence of misuse of genetic information in other jurisdictions is certainly indicative of similar potential misuse in the EU.

In accordance with this submission, it is also noted that the documented history of eugenics and abuse of science in Europe, as discussed in chapter 3, is a further indication of the potential future application of new genetic science. This points to a strong need to regulate this area.

6.2 Scope of Article 19

An initial concern that arose in the context of considering the legal basis for action in this area (which arguably has been ameliorated with the recent case law of the European courts) is the traditionally narrow and restrictive interpretation of the disability ground taken in the EU.¹³³ Such a concern may have presented challenges in terms of adopting a wide interpretation of Article 19 to encompass discrimination on the grounds of genetic predisposition to disability. However, this position has been brought in line with modern thinking in disability discourse with the recent cases, noted above. The concept of disability in the EU has now evolved to reflect a social model definition and one that is in line with the CRPD. Accordingly, discrimination on the basis of genetic predisposition to disability (or genetic information is arguably covered under Article 19.

¹³³ Case C-13/05 *Chacon Navas v Eurest Colectividades SA* [2006] ECR I-6467

6.3 The principle of subsidiarity and proportionality

Another potential challenge to EU level action in this area relates to the principle of subsidiarity. As noted, EU level legislative action is only necessary in circumstances where Member States are not able to regulate effectively. This principle provokes consideration of the existing legislative endeavours in the EU Member States. As has been highlighted, some Member States have already enacted legislation in this area, which has been specifically tailored to their legal systems and national traditions. The question arises whether the Member States can address this area more effectively. It is noted that EU level regulation may adversely impact upon local traditions in relation to insurance and employment regulation.¹³⁴

However, it is also arguable that a patchwork of protection offers incomplete and unequal protection and results in practical burdens for insurance companies and employers operating in different Member States. It is argued that EU level action in this area would operate to remove the existing barriers in the EU and would facilitate the accessibility of goods, services and people in this area. Further, in an age of advancing genetic technology, the potential for misuse will become more evident, and the need for comprehensive legislation more apparent. The principle of subsidiarity is therefore addressed. Similarly, the principle of proportionality arises and in response to this it is noted that the proposed genetic information non-discrimination directive does not endeavour to go beyond what is necessary in order to achieve its purpose and aims. The proposed legislation is narrow, discrete and focused on controlling the discriminatory use of genetic information in employment and insurance.

6.4 The segregation of genetic information

Another potential challenge that may be faced concerns the proposal to segregate genetic information (from other types of personal information) in a stand-alone directive. To a large extent, this argument has been addressed in chapter 5 and the discussion on whether to pursue a stand-alone mode of regulation. It proposed that stand-alone regulation is the preferable response in this area. The segregation of the ground of race and ethnic origin attracted criticism as discussed in chapter 9. The question is provoked whether a directive tailored to the ground of genetic information might also attract similar criticism. There will of course be diverging viewpoints on the proposal to segregate genetic information, however, on balance, it is submitted that such an approach will most effectively address the use of genetic information in EU. This model would offer clarity and certainty and would offer the opportunity to tailor specific protections to the issues arising and recognise the competing interests.

¹³⁴ Janneke H. Gerards and Heleen L. Janssen, 'Regulation of Genetic and Other Health Information in a Comparative Perspective' (2006) 13 *European Journal of Health Law* 339, 373

7. Options for regulation

Having put forward a *prima facie* case for EU level regulation and the merits of such an approach and having set out a clear dual legal basis for EU level action, this section will examine the options available for such action and will consider what shape this legislation might take. It will also consider the scope and content of such legislation.

On considering the options for EU level regulation of this area, reference is made to chapter 5, which discusses the mode of regulation and choice of regulatory frameworks. As noted, the focus of regulation is on non- discrimination. The objective of EU level regulation in this area should be to fully protect individuals against the discriminatory use of genetic information. In terms of mode of regulation, it is preferable to adopt a stand- alone approach, as highlighted in chapter 5.

7.1 What shape might an EU level response take

The next question that falls to be considered is whether it is preferable to introduce such a regulatory regime by way of a directive or a regulation. Regulations are an effective and powerful form of EU level legislation. It is the most direct form of EU law and is directly incorporated into the legal systems of Member States. The Member States do not have to take any independent action in order to transpose such laws in national legal systems. In comparison, directives require active measures by Member States in order to implement the law in the national legal systems.¹³⁵ The majority of EU legislation in the area of non- discrimination takes the form of directives.

It is submitted that a preferable means of addressing this area is by way of a directive. It is a more flexible instrument. Such a form of regulation would create a uniform standard at EU level and will facilitate the required objectives, while at the same time allowing for national traditions and norms in terms of transposing the legislation into national legal systems.¹³⁶ In consideration of the sensitive nature of this area and the historic sensitivity of previous abuses of science and eugenics in certain EU jurisdictions,¹³⁷ there may be further merit for allowing some leeway for national level implementation, while at the same time creating a certain harmonised standard at EU level. Further, it is noted that the previous and existing EU non- discrimination framework has illustrated that a directive is the preferable instrument, as highlighted in chapter 9.

¹³⁵ John Fairhurst, *Law of the European Union* (6th edn, Pearson Education Limited 2007) 62

¹³⁶ *Ibid* at 62

¹³⁷ Refer to chapter 3

7.2 Material scope of the proposed directive

The following section will consider the material scope and content of the proposed directive. As highlighted, this thesis has focused on the contexts of employment and insurance. In the US, the focus of the legislation was also on employment and (health) insurance, particularly in recognition of the clear connection between employment and health insurance in the US. In Australia, the main focus has been primarily in the areas of employment and insurance. It is also noted that many national level regulatory responses have similarly singled out the employment and insurance fields.

As noted, it is submitted that employment and insurance represent important gateways to accessing other necessary social goods and services, as well as facilitating an individual's full and active participation in society. In addition, from a disability perspective, it is observed that the creation of further barriers to employment or insurance may have a particularly adverse effect on persons with (putative) disabilities and other vulnerable members of society such as elderly persons. Therefore, as regards the scope of this proposed genetic information non- discrimination directive, it is submitted that it should extend to cover the areas of employment and insurance.

On consideration of the existing non- discrimination framework, as discussed in chapter 9, reference is made to the previously enacted non- discrimination directives. With a view to maintaining some degree of uniformity as regards the language used in these directives, the proposed genetic information non- discrimination directive should adopt similar language used in the existing directives. On considering the scope and content of the proposed legislation, particular reference is made to the Race Directive, as an illustrative comparison. The Race Directive has a broad scope of protection against discrimination on the grounds of race or ethnic origin.

7.3 Content of proposed directive

This section will set out the content and objective for the proposed genetic information non- discrimination directive. Firstly, the purpose of the directive is to provide a framework for combating discrimination on the grounds of genetic information, with a view to putting into effect in the Member States the principle of equal treatment. It provides a uniform minimum level of protection for individuals in the EU.

It is necessary to propose a definition of discrimination, for the purposes of the proposed directive. In line with the existing non- discrimination directives, the principle of equal treatment shall mean that there shall be no direct or indirect discrimination on the grounds of genetic information.

- (a) Direct discrimination shall be taken to occur when one person is treated less favourably than another is, has been or would be treated in a comparable situation on grounds of genetic information.
- (b) Indirect discrimination shall be taken to occur where an apparently neutral provision, criterion or practice would put persons with a certain genetic information at a particular disadvantage, compared with other persons, unless that provision, criterion or practice is objectively justified by a legitimate aim and the means of achieving that aim are appropriate and necessary.

It is also necessary to clarify the definition of genetic information, or genetic data. For the purpose of the proposed directive, genetic data shall mean: *“all data, of whatever type, concerning the characteristics of an individual that are inherited or acquired during early prenatal development.”*¹³⁸ It is noted that this definition mirrors the proposed definition of ‘genetic data’ in the draft Data Protection Regulations. In light of the submission made in this thesis that the current data protection reform would be supplemented and complemented by a proposed genetic non- discrimination directive, it is prudent to ensure uniformity in the definitions, as far as possible.

In terms of scope, this proposed directive shall apply to all persons, as regards both the public and private sectors, including public bodies, in relation to:

- (a) conditions for access to employment, to self- employment and to occupation, including selection criteria and promotion;
- (b) employment and working conditions, including dismissals and pay;
- (c) access to and supply of insurance services which are supplied to the public, as regards both the public and private sectors.

As regards the content of the proposed Directive, it is submitted that further research, discussion and consultation with the relevant stakeholders is required.

7.4 Exceptions

With a view to acknowledging the competing rights in this area, it is submitted that there is a need to include a number of exceptions in this proposed legislation. Chapter 3 examined third party interests in genetic information. It highlighted the competing interests and the limited right to know of both employers and insurers. In the employment context, it asserted that employers have a limited right to know about employees’ genetic information for

¹³⁸ Regulation of the European Parliament and of the Council on the protection of individuals with regard to the processing of personal data and on the free movement of such data (General Data Protection Regulation) COM (2012) 11 final, Article 4 (10)

occupational health and safety objectives in hazardous work environments (albeit under strict conditions). A similar exception has been provided for in the US, as highlighted in chapter 7. The German legislation in this area also contains a similar exception,¹³⁹ as does the Portuguese legislation.¹⁴⁰ Further, as noted in chapter 6, Article 27 of the CRPD can be interpreted as recognising an employer's right to know, in certain circumstances.¹⁴¹

In Australia, the Law Reform Commission (ALRC) recognised the limited right to use genetic information for occupational health and safety reasons.¹⁴² The ALRC made a number of observations in ascertaining the correct use of genetic technologies in these circumstances. It referred to the evidence of a clear link between the environment and the development of the condition, the condition in question may significantly harm the health or safety of employees and the genetic technology must be scientifically reliable and valid.¹⁴³

Therefore, on consideration of the limited right to know of employers, and in acknowledgment of the comparative perspectives, it is recommended that the proposed directive include a provision that recognises the right of employers to use genetic information, for occupational health and safety reasons. In these circumstances, the genetic information obtained is only to be used to monitor the biological effects of toxic or otherwise hazardous substances or activities in the workplace, with a view to detecting genetic based illness, injury or health condition which may develop in conjunction with certain workplace substances or activities. However, there is also a need for a number of safeguards. Such use of genetic information should also be the least intrusive means of carrying out the assessment in question. There must be a strong likelihood that the illness, injury or health condition will occur as a result of the workplace or employment activity in question.

The need to consider the scientific reliability and accuracy of the genetic information is acknowledged in this thesis. It is submitted that any use of genetic information in both employment (and insurance) be scientifically reliable and accurate. Otherwise, it is acknowledged that there is a potential for misinterpretation of the information and misuse.

On permitting such use of genetic information, it is also necessary to ensure that the information is obtained voluntarily and that the consent of the employee/potential employee is obtained. It is important that the employer maintain the privacy and confidentiality of the information at all times, in accordance with data protection obligations. It is also important to ensure that the information is not

¹³⁹ Article 20

¹⁴⁰ Article 13

¹⁴¹ UN CRPD, Article 27

¹⁴² ALRC Report, Recommendation 32

¹⁴³ ALRC Report, Recommendation 32-1

used in a discriminatory manner. It is noted that the exceptions in GINA in the employment context are framed in a similar manner, as noted in chapter 7.

There is also a need to consider the right to know of insurance companies. On acknowledgement of the economic viability of the life insurance industry and the risk of adverse selection, (as discussed in chapter 3), a narrow exception to the prohibition on the use of genetic information by insurers should be considered. It is submitted that life insurers should be allowed to use genetic test results (from tests previously taken by insureds), only in circumstances where the insurance cover sought is above a certain amount. In terms of the amount above which such information can be sought, it is acknowledged that further research and consultation is needed here. In consideration of such a provision, it is noted that the German legislation stipulates €300,000 above which genetic information can be sought in life insurance contracts.¹⁴⁴ In the UK, the moratorium states that £500,000 is the amount above which certain test results can be used, in life insurance contracts.¹⁴⁵

On permitting such use of genetic information, there is a need for a number of safeguards. It is necessary to ensure that the information is disclosed voluntarily by the insured and that the consent of the insured is obtained. It is also submitted that only genetic test results that are confirmed as being statistically valid and accurate are used in these circumstances. As regards the content and scope of this exception it is noted that further research, discussion and analysis is required with the relevant stakeholders in the area.

8. Can legislation alone solve the problem?

In light of the complexity of this issue and in consideration of the speed at which the technology is advancing, the question arises whether legislation alone can solve the problem. It is acknowledged that there is a real potential for a lack of awareness of the realities and limitations of advancing genetic science, as well as a lack of awareness of the existence of legal protections.¹⁴⁶ In light of the importance of ensuring that public confidence in this area is given paramount consideration, the following section will discuss additional means of addressing this issue.

8.1 Awareness raising

¹⁴⁴ Human Genetic Examination Act (Genetic Diagnosis Act GenDG) 374/09, 2009, section 4, article 18

¹⁴⁵ Concordat and Moratorium on Genetics and Insurance, para 25

¹⁴⁶ John Durant et al, 'Public understanding of the new genetics' in Theresa Marteau and Martin Richards (eds) *The Troubled Helix: Social and Psychological Implications of the New Human Genetics* (Cambridge University Press 2000, first published in 1996)

Studies carried out on the issue of genetic literacy have indicated a lack of awareness amongst the public of genetics and what it means.¹⁴⁷ Reference is also made to studies that took place in the US indicating that a large number of individuals were still concerned about the misuse of genetic information. This lack of awareness of the legal protections was also evident amongst the physicians surveyed.¹⁴⁸ In Australia, earlier studies also highlighted a concern for the issues arising as science and technology continue to advance.¹⁴⁹ In this regard, reference is also made to studies discussed above indicating fear of discrimination.

This points to the need for some form of awareness raising campaign to operate in conjunction with legislation, to ensure that individuals are aware, not only of the basic elements of genetic science but also the potential for misuse of their genetic information. Further, it is important to inform as to the legal protections in place. This awareness raising campaign should also target medical professionals and ensure that they are educated as to the legal issues that may arise with use of genetic information, as well as the protections in place. Therefore it is submitted that there is a need for both an awareness raising and an educational campaign. Awareness raising and education is key for facilitating and enhancing public trust and confidence in science and technology in the EU, which is a key objective in this area, as discussed above.

In support of this submission, it is noted that a number of international legal instruments similarly advocate for such awareness raising and education, including Article 8 of the CRPD.¹⁵⁰ In addition, some of the UNESCO soft law instruments provide for bioethics education, training and information, with a view to increasing knowledge of genetics and fostering public engagement.¹⁵¹

In the US, it is also noted that there has been recognition of the need for such awareness raising campaigns. A non- profit, online resource comprised of Genetic Alliance, the Genetics and Public Policy Center at Johns Hopkins University, and the National Coalition for Health Professional Education in Genetics was established in 2010.¹⁵² This resource aims to disseminate information about the legal protections available in the US. In addition, the

¹⁴⁷ Angela D. Lainie et al, 'Exploring the Public Understanding of Basic Genetic Concepts' (2004) 13 Journal of Genetic Counseling 4 305. See also Rafael Pardo et al, 'Attitudes toward biotechnology in the European Union' (2000) 98 Journal of Biotechnology 9

¹⁴⁸ 5th Cogent Genomics, Attitudes and Trends study, conducted by Cogent Research, Cambridge, Massachusetts (January 2011). See also Samuel W. Anderson, 'Genetic Privacy Worries on the Rise' – Interview with Christy White, Cogent Research, (2011) 24 Gene Watch 1 15. See additional studies referred to in chapter 7.

¹⁴⁹ Millward Brown Australia, Biotechnology Public Awareness Survey Final Report, Biotechnology Australia (21 August 2002). Available at www.biotechnology.gov.au/MBsurveyresults.pdf (accessed 25 July 2013)

¹⁵⁰ Refer to chapter 6

¹⁵¹ International Declaration on Human Genetic Data 2003, Article 24; Universal Declaration on Bioethics and Human Rights, Article 25

¹⁵² See <http://www.ginahelp.org/> (accessed 25 July 2013)

American Civil Liberties Union initiated a campaign, 'Take Your Genes Back', which aimed to raise awareness of the Myriad gene patent case, and its implications.¹⁵³

8.2 Human genetics advisory network

Linked to the previous argument and in light of the rapid pace of scientific advances and the inevitable potential for new forms of misuse of genetic information, it is submitted that there is a need for a human genetics advisory network. This network would monitor scientific and technological advances, address the new ethical and legal issues and challenges that may emerge, and advise the EU accordingly. It would also review the legislation periodically, with a view to monitoring its effectiveness. It is also acknowledged that upon introduction of a directive, the Commission appoints an expert group to assess the transposition of the Directive, engage in thematic research and advise the Commission. However, an additional advisory network may be necessary in light of the complexity and sensitivity of the area, as well as the inevitable potential for new challenges to arise. As noted in chapter 7, similar advisory bodies have also been recommended in the US under GINA¹⁵⁴ and in Australia, as recommended by the ALCR.¹⁵⁵ The recommendation for the establishment of such an advisory body in this area is also reflected in number of international instruments.¹⁵⁶

It is noted that there are a number of EU advisory bodies that may be in a position to fulfill this role. The European Commission's Group on Ethics in Science and New Technologies is a multidisciplinary body that reports directly to the Commission on matters of science and new technologies, and the ethical issues arising.¹⁵⁷ This group is arguably well placed to advise on new and emerging issues arising in this area. It has previously issued opinions in the area of genetic technologies.¹⁵⁸

From a human rights perspective, it is submitted that the Fundamental Rights Agency of the EU (FRA) may also be appropriate to take on this role.¹⁵⁹ In terms of the remit of FRA's activities, it is acknowledged that the organisation is tightly

¹⁵³ See <http://www.aclu.org/take-back-your-genes> (accessed 25 July 2013)

¹⁵⁴ GINA, s 208 (b)

¹⁵⁵ For further details, see <http://www.nhmrc.gov.au/about/committees-nhmrc/human-genetics-advisory-committee-hgac> (accessed 25 July 2013)

¹⁵⁶ For example, see Universal Declaration on the Human Genome and Human Rights, 1997, Article 24; International Declaration on Human Genetic Data 2003, Article 26; Universal Declaration on Bioethics and Human Rights 2005, Article 19; United Nations Convention on the Rights of Persons with Disabilities, Article 33

¹⁵⁷ Communication establishing the European Group on Ethics in Science and New Technologies (EGE) SEC (7) 2404

¹⁵⁸ Opinion No 18 of the European Group on Ethics in Science and New Technologies: Ethical aspects of genetic testing in the workplace (28/07/2003)

¹⁵⁹ Council Regulation (EC) No 168/2007 of 15 February 2007 establishing the Fundamental Rights Agency

constrained as regards its work programme, which is dictated by the European Commission. However, it is also pointed out that FRA is already active in the areas of non-discrimination and data protection. It is also acknowledged that by virtue of the EU Charter of Fundamental Rights, the areas of genetic discrimination and genetic privacy are areas that come within the scope of FRA's activities.

In addition to the above bodies, there are a number of other EU authorities that could potentially also have a role to play in this advisory network. The European Insurance and Occupational Pensions Authority (EIOPA), as an advisory body to the European Parliament and the Council, would be well placed to contribute from the perspective of the insurance context. The European Agency for Safety and Health at Work, which focuses on occupational health and safety, may be an appropriate body to contribute, from the perspective of employment. The inclusion of these bodies in the proposed network would ensure that the competing interests in this area are acknowledged. It is also proposed that the European Ombudsman should have a role to play in this network, to acknowledge the public interest perspective. From a data protection perspective, it is proposed that the Article 29 Data Protection Working Party have a role to play in this network. The inclusion of this group may also facilitate consistency between the EU data protection framework and the proposed genetic non-discrimination directive.

It is submitted that the European Group on Ethics in Science and New Technologies, FRA, EIOPA, the European Agency for Safety and Health at Work, the Article 29 Data Protection Working Party and the European Ombudsman are well placed to address this area from an advisory capacity. These bodies would provide a multifaceted perspective towards monitoring the proposed legislation, anticipating future challenges and representing the competing interests arising.

9. Conclusion

On building the case for EU level action in this area, the first part of this chapter highlighted the diversity of approaches in the EU Member States, with the conclusion that there is no harmony between the different jurisdictions. Such a patchwork of protection forms the basis for an argument in favour of EU level action in this area. It is acknowledged that such a patchwork has a negative impact on the EU internal market, including the economic activity and integration of persons with putative disabilities.

An additional argument for an EU level response is put forward in this chapter, which has as its objective the need to enhance public trust and confidence in science and technology. It is submitted that a preemptive EU level response is necessary in order to fully protect EU citizens against the misuse of genetic information and to instill confidence in individuals of the benefits of genetic science and technology.

This chapter proposes the case for an EU level non- discrimination approach. It acknowledges the positive developments in EU data protection law and it asserts that the proposed data protection framework acts as a practical starting point for legislating in this area. It is submitted that a proposed genetic non- discrimination directive will complement the new data protection framework.

This chapter submits that there is a clear legal basis in Article 19 TFEU and Article 114 TFEU. It proposes that a dual legal basis is appropriate here, to reflect the EU objective of addressing discrimination, as well as ensuring the functioning of the internal market. It reflects the growing cohesion between the economic objectives of the EU and the human rights objectives. In building a case for an EU level response, this chapter outlined the evidence of misuse of genetic information in the EU and other jurisdictions, with the objective of anticipating abuse in the future. The lack of firm evidence in the EU indicates the need for empirical research in this area.

This chapter also highlights the potential obstacles to such EU level action in this area. There is an evident gap in evidence and this needs to be addressed. Nevertheless, even in the absence of widespread evidence of genetic discrimination, this thesis considers the necessity of an EU level approach in anticipation of the potential for misuse. It was initially anticipated that challenges may arise in terms of the interpretation of Article 19; however, this position has been ameliorated by recent developments in the area of disability discrimination. This chapter also addressed potential concerns as regards the principles of subsidiarity and proportionality.

On consideration of the content of this legislation, reference is made to the existing EU non- discrimination framework. Accordingly, it is proposed the language used in the proposed directive reflect that in the existing non- discrimination directives. As regards the scope of the legislation, it recommends that it cover the fields of employment and insurance. The proposed legislation includes a number of exceptions, with a view to acknowledging the competing rights in employment and insurance. It recognises the limited right to know of employers and insurers.

Finally, this chapter concludes that legislation alone is not likely to be sufficient to adequately address this area, protect genetic information and enhance public confidence. It proposes the need to consider an awareness raising and educational campaign. It also found that there is a need to consider the establishment of a human genetics advisory body or network, with a view to monitoring scientific and technological developments, periodically reviewing the legislation and anticipating future legislative and policy challenges in this area.

Chapter 11: Conclusions and recommendations

1. Conclusions

Against the backdrop of rapid scientific and technological developments, the main objective in this thesis was to address the need for and the options available for an EU level regulatory response to address and control the use of genetic information by third parties, namely employers and insurance companies. On addressing this question, the following section will present the conclusions, which will be divided into four parts and will offer evaluations based on the findings and submissions of this thesis. This chapter will then present the recommendations proposed, including the legislative proposals at EU level.

1.1 Part one

Chapter two of this thesis found that genetic science and technology is advancing rapidly, with the result that genetic testing technologies are becoming more sophisticated. Genetic information is therefore becoming more available and accessible to individuals and to interested third parties, for medical and non-medical purposes. This chapter provided a number of insights into the potential as well as the limitations of genetic science and technology. Scientific and technological advances are revealing the genetic basis of a wide range of disease and disability, as well as behaviour and personality traits. It offers the potential of predicting the onset and potential severity of future disability and disease.

In terms of limitations, this chapter highlighted the current limited accuracy of most genetic technologies, which is a key factor to note when considering third party use of genetic information and a key factor in considering the regulation of genetic information. It is also noted that the accuracy of genetic technologies is likely to improve with future advances. On anticipation of the future orientation of genetic science, it is likely that new technologies will further advance and will be used more routinely in health care, with the potential to enhance public health for all.

Against this scientific backdrop, chapter three extrapolated the key ethical and legal issues as well as public policy concerns arising as a result of scientific and technological advances. A clear conflict of rights arises between the individual, family members and third parties. The recognition of the public policy concerns offers an insight into the potential consequences if this area is inadequately regulated. Although there are no clear answers to these ethical issues and public policy concerns, the relevance of this chapter was to provide an ethical perspective to this discussion.

Chapter three concentrated primarily on third party interest in genetic information. On considering third party use of genetic information, this chapter narrowed the focus to the employment and insurance contexts, in light of the fact that employment and insurance are both portals to accessing a myriad of social and economic goods and services, thereby impacting upon an individual's private and professional life, as well as participation in society.

In the employment context, it illustrates the conflict of rights between the employee and the employer. This chapter submits that an employer has a limited right to know in certain circumstances, albeit under strict conditions, and in recognition of the limited accuracy of genetic technologies. The reconciliation of these rights may be achieved by ensuring that the area is appropriately regulated, taking account of the rights of employees to privacy and not to be discriminated against, and the limited right to know of employers in certain workplace scenarios.

This chapter similarly highlights the conflict of rights that arises between the insurer and the insured. This chapter found that an insurer's interest in genetic information stems from the fundamental principles and practices of insurance law, as well as from a concern for the economic objectives of the insurance industry. Similarly, an insured has the right not to be discriminated against and a right to privacy, as well as a right to access insurance.

In acknowledging the competing rights, this chapter asserts that the use of genetic information by insurers must be regulated, while at the same time recognising the economic objectives of insurance companies. In support of an insured's rights, particular reference is made to the recent *Test Achats* case, which signals an expansion of non-discrimination principles in the insurance field in the EU. Reference is also made to the EU Charter of Fundamental Rights and the protection against discrimination on grounds of 'genetic features.'

With a view to upholding an individual's right to autonomy, right to privacy and right not to be discriminated against, insurers shall not request that insureds undergo genetic testing under any circumstances. It is submitted that insurers should not request family history information under any circumstances. It is also submitted that insurance companies shall not request the results of genetic tests previously taken, except in respect of life insurance contracts above a certain amount (the exact amount of which requires further consideration), and only in circumstances where the results of the genetic test can be confirmed as being statistically valid and accurate.

These submissions recognise the need for a compromise in acknowledging the economic viability of the insurance industry. The proposal of a ceiling amount (below which no genetic information can be requested) ensures that life insurance still remains accessible to all, and at the same time it may prevent the

abusive use of genetic information by customers and ameliorate the possibility of adverse selection. By requiring confirmation of the statistical validity and accuracy of genetic test results, this may also ensure that genetic information is not misinterpreted or its value overestimated.

Although this thesis looks primarily at the products of health insurance and life insurance (as it is submitted that these are the most immediately relevant in the area of private insurance), it is acknowledged that further types of insurance products may require consideration as regards the use of genetic information. This will require further research and discussion in the future with all interested stakeholders.

Chapter three emphasises that any potential use of genetic information by third parties must consider the limited accuracy of most genetic technologies. This also provokes consideration of the need for some form of educational and awareness raising campaign to inform as to the realities of genetic science and technology.

Chapter four highlights the application of the social model of disability to the evolution of genetic science. This chapter adopts the theory of the social model to address the challenging issues that arise at the intersection of advancing science, ethics and the law. With a focus on the societal, attitudinal and other external barriers that disable an individual, it provides a core human rights based model and concentrates on acknowledging and achieving the rights of persons with putative disabilities.

Part one of this thesis provided a clear background framework from which to consider the issue of regulation. By highlighting the concerns arising at the intersection of advancing science, technology and ethics, it facilitated a more focused discussion of this topic and presented a clear picture indicating the need for regulation in this area.

1.2 Part two

Part two of this thesis focused on the regulation of genetic information. As regards the mode of regulation, it asserts that a stand-alone model that singles out genetic information is the most appropriate approach. This approach recognises that genetic information is special, has unique characteristics and therefore merits special protection. In light of the history of eugenics and genetic cleansing which was evident in Europe, it is submitted that such an approach would make an impact from a political and moral perspective in terms of controlling the use of genetic information. Chapter five also highlighted a preference for a hard law approach, based on the preference for certainty, strong enforcement mechanisms and expressive value.

In terms of the choice of regulatory frameworks in legal theory, chapter five examined the non- discrimination approach, the privacy approach (which finds expression in the data protection model) and the property approach. It found that the property framework is inappropriate in these circumstances, particularly in light of the potential for further commodification of genetic information. With a view to ensuring the full protection against misuse of genetic information, this chapter found that it is preferable to consider both a non- discrimination approach and a privacy/ data protection approach. Such a two- pronged model would ensure that access to and disclosure of genetic information is controlled, and that the discriminatory use of genetic information is prohibited. The conclusions in chapter five provide the basis for discussion of the international and comparative benchmarks in this area, as well as the current framework in the EU.

Chapter six highlights the imperative for a regulatory regime in this area from an international human rights law perspective. It highlights the evolution in the interpretation of human rights as science and technology advance, and as societal norms change, with reference to the relevant UN treaty law, as well as the soft law instruments. It is clear that at international level, there is an awareness of the potential of advancing science and technology and the impact on fundamental human rights. This is particularly evident in the UN soft law instruments which specifically address the area of bioethics and human genetics.

In line with the social model of disability paradigm, it is submitted that the CRPD encompasses discrimination on the basis of genetic predisposition to disability, therefore covering individuals with putative disabilities. It found that the CRPD, as a key disability rights instrument, has the potential to drive law reform in this area. The conclusion of the CRPD by the EU is a new departure in the area of human rights in the EU, illustrating the commitment of the EU to addressing these issues. This chapter noted the potential of this development to provoke action in this area. From this analysis of the international human rights framework, it is clear that the UN instruments provide a clear moral impulse for regulation of this area.

Chapter seven examines evolving comparative law benchmarks and focuses on the US and Australia, which are two of the most developed jurisdictions from a legislative and policy perspective. The US is arguably a pioneer in this area, and has introduced federal level legislation, specifically tailored to the regulation of genetic information. There is much to be learned from the US approach. GINA is a preemptive law, introduced with a view to enhancing public confidence, anticipating a type of discrimination that is likely to accelerate in the future, as science and technology continue to advance. The legislation was also needed in consideration of the history of eugenics and misuse of science in the US. Such history of eugenics is also evident in Europe, and may provide the historical context to compel legislation in this area in the EU. Similarly, there is also an absence of evidence in the EU in this area. In addition, GINA was also introduced to address the unsatisfactory patchwork of laws at state level. This is

comparable to the current patchwork of laws that exists in the EU Member States.

Similarly, there is much to be gained from examining the Australian position. The empirical research carried out at national level is admirable and provides significant insights into the reality of genetic discrimination (and other misuse of genetic information). In consideration of the evident gap in research in the EU in this area, the Australian model provides a valuable example. This major project was complemented by an extensive investigation carried out by the ALRC. In contrast to the US, Australia rejected the option of a new regulatory regime (in favour of amending existing legislation and policy) and viewed the issue from both a non-discrimination and a privacy perspective. The ALRC report provides an in-depth analysis of this area and a valuable point of reference for future legislative endeavours in the EU.

1.3 Part three

The objective of part three was to examine the European perspective. Chapter eight provided an overview of the current human rights norms in the Council of Europe (CoE) and the regulation of genetic information. It documents the CoE position, in providing a moral framework from which to view this area and illustrates how the relevant instruments can shape our understanding of the ethical and legal issues arising from advancing genetic science.

There is a growing awareness in the CoE of scientific and technological developments, and the challenges presented. The ECHR provides a strong human rights framework and the case law of the ECtHR has recognised the privacy of genetic information, as well as discrimination on the basis of genetic predisposition to disability. The Biomedicine Convention (and its Protocol) provides further guidance on these issues, from a moral perspective. This chapter found that several soft law instruments of the CoE have also addressed this area and offer guidance. The CoE has acknowledged the need to respond to scientific and technological advances and develop a regulatory response based on a human rights framework.

Chapter nine focused on the regulatory position in the EU and how it might apply to the regulation of genetic information. On building upon the findings of chapter eight, this chapter highlighted the human rights framework in the EU, with a focus on the CFREU. It found that the elevated status of the CFREU, and the specific reference to discrimination on the basis of 'genetic features' places this area in a firm human rights position within the EU legal framework. It is submitted that this instrument can influence the proposal of legislation in this area. Although it is acknowledged that the Charter does not afford an additional legal basis, it certainly acts as an impetus to action in this area.

Chapter nine highlighted the strong data protection framework in the EU. The intention to update data protection laws will have the effect of controlling the access to and disclosure of 'genetic data' in the EU. This is a welcome development and a key legislative starting point in this area. This updated data protection framework will therefore be complemented by any subsequent legislative proposals at EU level.

Chapter nine explored the reach of current EU non- discrimination law and how it might apply to the regulation of genetic information, particularly through an interpretation of the disability ground. It found that there is a strong non-discrimination framework in the EU. Importantly, the concept of disability in the EU has evolved from a restrictive medical model approach to a more modern construction based on the social model of disability and in line with the CRPD. Certain gaps were identified in the non- discrimination framework that would point towards the need for further protections to comprehensively cover discrimination on the basis of genetic predisposition to disability in the areas of employment and insurance. Chapter nine therefore indicates the need to consider a new regulatory framework in the area of non- discrimination.

1.4 Part four

In drawing together the conclusions in parts one, two and three, chapter ten builds a case for EU level action in this area. It documents the significant diversity in the EU MS responses in this area. This patchwork and the lack of uniform regulation is presented as a key argument indicating the need for EU level regulation in this area. In particular, it potentially creates problems for the functioning of the EU internal market. On building the case for an EU level response, chapter 10 also demonstrates the need to enhance public confidence in advancing genetic science and technology. This argument is important from the perspective of improving public health and driving economic growth in the EU. It is submitted that in order for individuals to be fully confident that their genetic information is protected, and in order to encourage individuals to take advantage of genetic technologies, it is necessary to have in place an appropriate regulatory regime.

The key proposal made in this thesis is therefore the need for an EU level response to address the discriminatory use of genetic information. In support of such a proposal, reference is made to the CRPD and the CFREU, both of which act as an impetus to action in this area. In making this submission, it is emphasised that the current data protection reform process that is taking place in the EU will be complemented by the proposed directive on genetic non-discrimination.

Chapter 10 considers a dual legal basis for EU action in this area. The submission for a dual legal basis is made in recognition of the evident re-orientation of EU priorities from solely economic objectives to incorporating both

economic objectives and human rights objectives. In making this submission, reference is made to the Council Decision to conclude the CRPD, which also relied on the dual legal basis of Article 19 TFEU and Article 114 TFEU. This approach would have the effect of addressing discrimination, facilitating the functioning of the internal market and acknowledging the importance of the economic contribution of persons with putative disabilities.

On formulating a case for EU level action, chapter ten examines existing evidence of misuse (and fear of misuse) of genetic information. It found that there is little evidence. It also demonstrated that the majority of evidence has been gathered in other jurisdictions such as the US and Australia. Although there is some evidence of misuse in the EU, it is clear that there is a gap in research in this regard. This points to the need for empirical research in the EU in this area, to verify cases of misuse of genetic information, including discrimination and fear of discrimination.

Chapter 10 recognises the potential challenges to EU action, particularly in light of the absence of clear empirical evidence in the EU of actual misuse of genetic information. Nevertheless, even in the absence of documented evidence of genetic discrimination taking place, this chapter proposes an EU level response in this area. Therefore the rationale of such legislation is to anticipate abuse in the future, with the objective of enhancing public confidence, as well as in consideration of the speed at which science and technology is advancing.

In terms of the shape of this legislation, it proposes a stand- alone directive to address the discriminatory use of genetic information in employment and insurance. On proposing such a directive, reference is made to the existing EU non- discrimination framework, in terms of scope, content and definitions. In particular, reference is made to the Race Directive, which also singled race out in a stand- alone directive.

This chapter also raised the question as to whether legislation alone is sufficient to adequately address this area. It submits that in light of the speed at which science and technology is advancing and the complexity of the issues arising there is a real potential for lack of awareness, not only of genetic science but also of the legal protections in place. These insights indicate the need for an awareness raising campaign to target third parties, the medical profession, as well as the general public. It proposes that there is also a need for a human genetics advisory network in this area, with the objective of monitoring the legislation introduced, monitoring developments in the area and anticipating potential future challenges in the area.

2. Recommendations

On evaluation of the findings made in this thesis and in consideration of the conclusions drawn in this chapter, the following section proposes a number of final recommendations.

2.1 EU level regulatory framework

In consideration of the speed at which science and technology is advancing, the potential for breach of fundamental human rights and the myriad of ethical and legal dilemmas arising, it is submitted that there is clear need for an appropriate regulatory framework to address the use and misuse of genetic information. In light of the current patchwork of legislative frameworks in the individual EU Member States, it is submitted that the appropriate means of protection is a directive at EU level. Such an approach would ensure a uniform, minimum set of standards.

This recommendation is also made with a view to enhancing public confidence in genetic science and technology, for the benefit of public health and to facilitate further technological innovation in the EU. This recommendation draws together the objectives of economic advancement, public health and recognition of fundamental human rights in the EU. In making this recommendation, it proposes that even in the absence of a documented history of genetic discrimination in the EU, the proposed legislation may act to reassure individuals that they can engage in genetic testing and take advantage of such technologies without the fear of discrimination or privacy concerns. It may also encourage individuals to participate in clinical trials and research, which is necessary for further scientific and technological development.

Further, it is submitted that in order for the EU to further progress a growing genomics industry, it is necessary to have in place a strong EU level regulatory framework.¹ Reference is also made here to the exponential growth of the genomics industry in the US in the past few years, which is arguably, in some way, attributable to the introduction of a clear federal level regulatory regime.² In support of an EU level response, particular reference is made to the conclusion of the CRPD by the EU and the elevated status of the CFREU in the EU, both of which are an impetus to action in this area.

This thesis recommends a stand-alone genetic information non-discrimination directive, on the basis of Article 19 TFEU and Article 114 TFEU. As highlighted, it is submitted that a non-discrimination model at EU level in this area would

¹ A recent report found that in 2011 the United States had the largest market share in the genomics industry, with Europe having the second largest share. See Report, Transparency Market Research, 'Genomics Market, Global Industry Analysis, Size, Share, Growth, Trends and Forecast' 2012- 2018 (6 August 2013)

² Battelle Memorial Institute, Report: 'The Impact of Genomics on the US Economy' (June 2013), available at: <http://www.battelle.org/docs/health-and-pharmaceutical/the-impact-of-genomics-on-the-u-s-economy-june-11-final.pdf?sfvrsn=0> (accessed 14 August 2013)

supplement the proposed new data protection framework and would have the effect of fully protecting individuals from misuse of genetic information.

The aim of the proposed directive is to provide a framework for combating discrimination on the grounds of genetic information, with a view to putting into effect in the Member States the principle of equal treatment. The proposed legislation would prohibit both direct and indirect discrimination on the grounds of genetic information. In terms of the statutory language contained therein, it is recommended that the proposed directive reflect the language contained in the existing EU non-discrimination framework, to ensure consistency. In terms of the definition of 'genetic data', it is recommended that this mirrors the definition of 'genetic data' proposed in the draft data protection regulations.

In terms of the scope, it is recommended that this directive cover the areas of employment and insurance. With a view to achieving an appropriate reconciliation of interests, this proposed directive should contain carefully worded provisions setting out the limited circumstances in which use of genetic information is permitted. In the employment context, employers should be permitted to use genetic information, only under limited circumstances, with the objective of detecting potential genetic conditions in certain hazardous environments, pursuant to health and safety obligations and with a view to upholding the duty of care to the employee.

In recognition of the economic viability of the life insurance industry and the need to preempt the potential for adverse selection, it is recommended that the results of genetic tests previously taken are only to be taken into account in respect of life insurance contracts above a certain amount (to be further considered). Further, it is recommended that such use of genetic test results only take place in circumstances where the results of the genetic test can be confirmed as being statistically valid and accurate.

It is recommended that further research is required into the use of genetic information in other insurance products in the EU. Although private health and life insurance are the areas where use of genetic information has been most contentious and has generated the most debate, it does not discount the concern surrounding use of genetic information in other insurance products.

On consideration of any use of genetic information in both the employment and insurance contexts, it is important to ensure the scientific reliability and accuracy of the information.

2.2 Human genetics advisory network

This thesis also recommends the establishment of a human genetics advisory body or network that would ensure the full effectiveness and implementation of the proposed legislation in the EU, as discussed in chapter 10. The objective of

this network would be to advise the EU on policy and implementation of the legislative measures. In light of the rapid advances in genetic science and technology, it is acknowledged that there is a need to monitor these advances with a view to evaluating the legislation in line with these scientific developments. This network would also carry out regular reviews of the proposed legislation to ensure that it is responding adequately to such scientific and technological developments. This network would also anticipate future legislative and policy challenges that may arise in this area, as science and technology advance.

In support of this recommendation, reference is made to similar advisory bodies have also been recommended in the US and in Australia, as recommended by the Australian Law Reform Commission.³ The recommendation for the establishment of an independent advisory body in this area also resonates with the provisions in some of the international instruments.⁴ Reference is also made to Article 33 of the CRPD, which provides for the establishment of a monitoring mechanism, as noted in chapter 6.

This thesis recommends the establishment of a multi- disciplinary advisory network of experts. In order to reflect the multi- faceted nature of this area and to acknowledge the competing interests at stake, it is recommended that the composition of the proposed network include (but not be limited to) the following bodies: the Fundamental Rights Agency of the European Union (FRA), the European Group on Ethics in Science and New Technologies, the European Insurance and Occupational Pensions Authority, the European Agency for Safety and Health at Work, the Article 29 Data Protection Working Party and the European Ombudsman.

2.3 Awareness raising and education

In addition to legislation, it is recommended that there is also a need to consider an awareness raising and educational campaign, to target medical professionals, commercial entities and the general public. It is submitted that there is a need to raise awareness and educate as to the benefits (and limitations) of genetic science and technology, the potential for misuse of such technology, as well as the legal protections in place. It has been shown that in the US that there is still a lack of awareness of the legal protections in place and individuals are still concerned about the misuse of genetic information, even following the introduction of federal level legislation.⁵

In support of this recommendation, it is noted that a number of international legal instruments similarly provide for such awareness raising and education.⁶ Article 8

³ See chapter 7

⁴ Universal Declaration on Bioethics and Human Rights 2005, Article 19

⁵ Refer to chapter 7

⁶ See International Declaration on Human Genetic Data 2003, Article 24, Universal Declaration on Bioethics and Human Rights 2005, Article 25

of the CRPD advocates in favour of awareness raising to generate positive perceptions of disability.⁷ As interpreted in chapter 6, this provision might therefore manifest in the form of educational and awareness raising campaigns, with a view to generating positive perceptions of disability, and genetic predispositions to disability. In addition, Article 24 of the International Declaration on Human Genetic Data, 2003 provides for ethics education, training and information, with a view to increasing knowledge of genetics and fostering public engagement.⁸ Article 25 of the Universal Declaration on Bioethics and Human Rights, 2005 also provides for bioethics education, training and information.⁹

It is recommended that the proposed human genetics advisory network would be the appropriate body to engage in awareness raising and education in the EU.

2.4 Empirical research

In light of the evident lack of evidence of misuse of genetic information in the EU, this thesis recommends the need to engage in an empirical research project to provide further details, insight and evidence of the incidence of genetic discrimination, breach of privacy and other misuse of genetic information. In support of this recommendation, reference is made to the major empirical study that was carried out in Australia. This study proved successful, and informed the research and recommendations of the ALRC in this area. In the EU, such research would inform and support any legislative and policy endeavours. With reference to the Australian study, it is recommended that the proposed empirical research project take a multifaceted approach to this area, and consider the perspective of all relevant stakeholders including the public and commercial entities.¹⁰

It is further recommended that the proposed human genetics advisory network would be the appropriate body to engage in such empirical research. Specifically, it is noted that FRA may be particularly well placed to engage in this research and that such work would come within the scope of FRA's work. FRA gathers data to present to the EU institutions when the EU is implementing law.

2.5 Other recommendations

This thesis recommends that there needs to be further, more in- depth and focused discussion and consultation with the various stakeholders involved, including employer organisations, insurance organisations, lawyers and policy makers. This further discussion is particularly necessary in the insurance industry, in light of the complexity of the issues arising, and with a view to

⁷ CRPD, Article 8

⁸ Article 24

⁹ Article 25

¹⁰ Refer to chapter 7

achieving the correct calibration of competing interests. There should also be a public consultation and engagement on these issues to ensure transparency and to gauge the public's attitude and perception of these issues. Such public engagement would be particularly effective in the insurance industry, particularly with a view to tackling transparency issues. This endeavour would also operate to improve public confidence in this area, in line with the objective of the above recommendations.

3. Final conclusion

This thesis has considered and proposed the need for an EU level response to regulate the flow and use of genetic information by employers and insurance companies, in light of advancing science and technology and in consideration of the fundamental human rights at stake and the competing commercial interests arising. In order for genetic science and technology to further progress for the benefit of all and in order for the EU to fully reap the economic benefits of this genetic revolution, public confidence in these new technologies needs to be assured and maintained.

An EU level response, in the form of a genetic information non- discrimination directive may go some way towards protecting genetic information against misuse and acknowledging the competing interests arising. In particular, such a response would improve the inclusion of persons with putative disabilities in terms of accessing employment and insurance in the EU. In looking forward, it would ensure that a genetic underclass does not become a reality. A clear EU level response would also contribute towards enhancing public confidence at an early stage, as well as facilitating further advances in genetic science and technology. From an economic perspective, the progression of science and technology are key to the growth of the EU economy.

It is acknowledged that the insights given and the proposals made in this thesis are just a first step in the longer journey towards effectively grappling with the myriad of regulatory challenges presented as the law struggles to deal with rapid scientific and technological advances. Accordingly, a major challenge of the 21st century will be to find new and innovative ways to try to close the gap between law and technology, acknowledge emerging legal and ethical concerns, while at the same time allowing science to progress for the benefit of all.

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