Health Canada Santé Canada

Health Policy
Working Paper Series

Genetic Testing for Late Onset Diseases: Current Research Practices and Analysis of Policy Development

Working Paper 01-02
September 2001

Canada

Our mission is to help the people of Canada
maintain and improve their health.
Health Canada

This publication can be made available in/on computer diskette/large print/audio cassette/braille, upon request.

For additional copies or for an address change, please contact:

Health Canada
Policy Research Communications Unit
Tel.: (613) 954-8559
Fax: (613) 954-0813
rmddinfo@hc-sc.gc.ca

It is available electronically at the following address:
http://www.hc-sc.gc.ca/iacb-dgiac/nhrdp/index.html

Ce document est aussi offert en français sous le titre :
Test Génétiques de Détection des Maladies à Déclenchement Tardif : Pratiques Actuelles en Matière de Recherche et Analyse de L'élaboration de Politiques

The opinions expressed in this publication are those of the authors and do not necessarily reflect the views of Health Canada.

The Working Paper Series (WPS) supports evidence-based decision making by highlighting and promoting Health Canada's policy research to a broad number of policy analysts and decision makers in Health Canada and its partners in the external research community. The WPS is part of a larger policy research communications program that includes the *Health Policy Research Bulletin*.

Cat. N° H13-5/01-2E
ISBN 0-662-31031-4

Genetic Testing for Late Onset Diseases: Current Research Practices and Analysis of Policy Development

Christine E. Jamieson
Concordia University

Author Biography

Dr. Christine Jamieson, BTh, MA, LTh, PhD (Ethics), is an assistant professor in the Department of Theological Studies at Concordia University in Montréal. She teaches courses relating to social and applied ethics, the work of philosopher and theologian, Bernard Lonergan, and the relationship between religion and culture. Her expertise is in the area of social ethics, with a particular emphasis on analyzing underlying structures that influence value and value judgement in culture and society. She has significant strengths in the areas of genetic technology and its impact on society, issues concerning violence against women, and the psychosocial dynamics of conflict. Dr. Jamieson is currently engaged in research to develop a framework of analysis that could serve as a basis for ethical deliberation in a pluralist context.

Résumé

Ce rapport est le premier d'une série commandée par la Division des politiques de la Direction des politiques de la santé et des communications de Santé Canada, dans le but de donner un aperçu de l'information de base sur les tests génétiques servant à détecter les maladies à déclenchement tardif (MDT). Il traite à la fois des grands enjeux des pratiques actuelles en matière de recherche et des principaux facteurs qui façonnent l'élaboration des politiques en puisant dans la documentation européenne et nord-américaine. Trois autres rapports proposent une analyse en profondeur de thèmes précis : enjeux de principes et de juridiction, enjeux médicaux et sociaux, et enjeux éthiques et psychologiques. Santé Canada publiera les rapports sur les enjeux de principes et de juridiction, et l'auteure publiera elle-même les deux autres.

Les tests génétiques de détection des MDT sont une sous-catégorie du vaste ensemble de tests génétiques. Alors que les tests génétiques mis au point pour les maladies apparaissant pendant l'enfance constituent souvent un outil de diagnostic, ceux qui ont pour but de détecter les maladies à déclenchement tardif ont une optique entièrement nouvelle, puisque *la détection précède d'un bon nombre d'années le déclenchement de la maladie*. Essentiellement, ils servent à déterminer si des personnes en santé présentent un risque faible ou élevé de développer une maladie quelconque dans un avenir plus ou moins lointain. Ils créent une nouvelle catégorie sociale, celle des « personnes qui ne sont pas encore malades ». Cette sous-catégorie de tests génétiques s'accompagne de nouveaux problèmes qui méritent une réflexion et une analyse attentives à bien des niveaux.

Le rapport se divise en trois parties. La première partie présente de l'information de base concernant la technologie génétique et situe dans ce contexte les tests de détection des MDT. La deuxième partie donne un aperçu des 15 principaux enjeux liés aux tests génétiques de détection des MDT. L'analyse qu'elle présente se fonde sur la documentation européenne et nord-américaine. Les enjeux retenus sont ceux que l'on retrouve de façon constante dans la documentation. La troisième partie analyse les facteurs sous-jacents qui façonnent actuellement l'élaboration et la mise en œuvre des politiques touchant les tests génétiques de détection des MDT en Europe et en Amérique du Nord. En règle générale, il existe une convergence de base, sur le plan général, sur les enjeux importants en Europe et en Amérique du Nord. Toutefois, on note une divergence dans l'analyse plus approfondie de ces enjeux. La troisième partie utilise deux outils d'analyse : l'évaluation de la technologie et une échelle de valeurs.

Abstract

This paper is the first in a series of articles commissioned by the Policy Division, Health Policy and Communications Branch, Health Canada, to provide background information on genetic testing for late onset diseases. It provides an overview of current issues relevant to genetic testing, and analyzes the major factors shaping policy recommendations by drawing on European and North American literature. Three subsequent papers provide in-depth analysis of specific themes: policy and jurisdictional issues, medical and social issues and ethical and psychological issues. The paper on policy and jurisdictional issue will also be published by Health Canada, but the other two papers will be published by the author.

Genetic testing for late onset diseases is one component of the larger field of genetic testing. Whereas DNA testing for diseases with onset in childhood is often synonymous with diagnosis, genetic testing for late onset diseases is completely new because it *provides testing that predates the onset of disease by many years*. Essentially, genetic testing for late onset diseases diagnoses healthy individuals with either a high or low risk of developing a disease at some time in the future. It creates a new social category: the not-yet-ill. This component of genetic testing presents new challenges that require careful thought and analysis on many levels.

This paper is divided into three parts. Part one provides background information on genetic technology, and situates genetic testing for late onset diseases within this background. Part two gives an overview of 15 key issues concerning late onset genetic testing. The literature from which this analysis is drawn covers Europe and North America. The issues were chosen for their constant and consistent presence in the literature. In general, there is basic convergence at a general level in Europe and North America on the key issues. However, there is divergence on how each jurisdiction works out the details. Part three analyzes the underlying factors that are shaping policy recommendations for and implementation of genetic testing for late onset diseases in Europe and North America. Part three draws on two tools of analysis: technology assessment and a scale of values.

Table of Contents

Introduction

Unlike other scientific research, the Human Genome Project (HGP) has operated, almost from the time of its inception, under intense international scrutiny from many perspectives on a multiplicity of issues. The issues encompass ethical, social, legal, psychological, religious and philosophical concerns. The imperative behind this close examination emerged from past experiences in which science and technology forged ahead with innovations in the almost complete absence of ethical and social inquiry. In the past, science and technology tended to be left to the experts. Now, much more than before, interested non-expert individuals and groups are involved. In response, governments are recognizing their obligation and responsibility to address the implications of new technologies from a diversity of perspectives.

Technological innovation has the uncanny ability to outrun our ability to comprehend and assess its current and future implications. Science and technology's agenda, and the progress ensuing from that agenda, challenge humanity in unprecedented ways. For example, developments in the biological sciences and reproductive technologies have the potential to transform the human species. This awesome human ability brings with it the recognition of a new form of responsibility. Responsibility here means more than mere accountability for the past; it refers also to our responsibility for the future. In the extreme, the future of humankind is at stake.

The intention behind science and technology has been, at least in theory, to create a better world. Certainly, the HGP can be viewed in large part as motivated by a desire to create a better world through a deeper understanding of the human person, of the genetic component of disease and of possible correctives to hereditary diseases. The caution, however, that accompanies the HGP is warranted. There is ample evidence that technological innovation, even when pursued for potential benefits, has led to disastrous results. Thus, the intense ethical and social scrutiny that has emerged in response to the HGP is an attempt to *think* about what we are doing. It is an important effort, at the very least, to parallel the technological advances with thought about the ethical, social, psychological and other implications. Particularly striking is the fact that traditional ethical and social concepts are often not able to address the concerns that technological innovation raises.

Genetic testing is a significant product of the HGP. Among other changes, genetic testing has increased our ability to understand and treat disease; it is altering our understanding of the causes of disease; it is creating new challenges in relation to the delivery of health care. Genetic testing for late onset diseases is an important component of genetic testing. Whereas DNA testing for diseases with onset in childhood is often synonymous with diagnosis, genetic testing for late onset diseases is completely new because it *provides testing that predates the onset of disease by many years*. Essentially, genetic testing for late onset diseases diagnoses healthy individuals with either a high or low risk of developing a disease at some time in the future. It thereby creates a new social category: the not-yet-ill.[1] This component of genetic testing presents new challenges that require careful thought and analysis on many levels.

Key issues involving genetic testing for late onset diseases concern both governments and the citizens who elect them. For example, who should be tested for late onset diseases? How can the quality of laboratory work be ensured? How much counselling is required before administering the test? Who should have access to test results besides the individual being tested and his or her physician? These questions and many more are subjects of this paper, which consists of three parts.

Part one provides background information on genetic testing for late onset diseases and considers this testing within the broader theme of genetic technology. Research aimed at identifying the origins of and conquering human disease developed from the more general study of human genetics. What are the larger issues at stake? What material contributes to our general understanding of genetic technology that will aid us in considering the specific focus of genetic testing for late onset diseases? The findings in this part of the paper provide relevant data for parts two and three. This background section also provides a glossary of terminology related to genetic testing.

Part two looks at specific issues that pertain to genetic testing for late onset diseases. Each issue is considered from an international perspective, through current literature from Europe and North America.

Drawing on the information in parts one and two, part three analyzes various factors pertaining to the underlying presuppositions that shape policy formation in relation to genetic testing for late onset diseases. In considering both the general ethical, social, legal and philosophical factors concerning genetic technology and the specific ethical, social, legal, cultural, psychological and other factors pertaining to genetic testing for late onset diseases, part three identifies presuppositions behind policy recommendations and policy implementation.

Background

Genetic Technology

To designate genetics as a technology is already to say something quite significant. Genetics is the branch of science that deals with heredity and the variation of inherited characteristics in living organisms. The word *genetics* is derived from the Greek word *génesis*, meaning generation, creation, nativity or horoscope.[2] Technology has to do with the work of human hands — the tools, machines, computers and factories of the Western world since the Industrial Revolution.[3] It is the branch of knowledge that deals with the mechanical arts or applied sciences. To juxtapose the word *genetic* with the word *technology* accounts for both the incredible excitement and the incredible fear that genetic technology generates. Paradoxically, both reactions stem from the same reality — our capacity to experiment with the origins of the human species and of every human person. Every aspect of genetic technology involves this "tinkering." The fundamental question concerns the difference between technology involving inanimate and non-human material on the one hand and technology involving human beings on the other. This is not a new question, since technologies involving human beings have existed for a long time. However, there is something unique about genetic technology, and this has given rise to massive international attention. As noted above, the attention is both positive (seeing the incredible potential that genetic technology will bring) and negative (a foreboding sense of the precariousness of genetic technology and the fear of potential disasters).

The well-known goal of the multinational HGP is to map and sequence the entire human genome. Mapping the human genome involves the classic reductionist method of science — that is, attempting to understand wholes such as organisms in terms of their ever-smaller parts, the cells and genes. The human genome comprises in its totality all the genes found in a human cell. Mapping refers to the assignment of human genes to locations on various chromosomes. Sequencing refers to the precise order of the bases (adenine, cytosine, guanine and thymine — designated A, G, C and T, respectively) that characterize the DNA of each gene. The benefits of molecular genetics in general and the HGP in particular for humanity are profound. "The increasingly powerful diagnostic, predictive, and life-enhancing tools generated by molecular genetics and biotechnology have already begun to revolutionize medicine."[4]

This new "power" brings with it significant challenges — challenges that encompass every aspect of human living. The social, ethical, psychological, religious and philosophical realms of human meaning are potentially affected by genetic technology. For example, one concern that is continually noted in international literature concerning genetic technology is the need for communication between the scientific and non-scientific realms. They represent different "worldviews" and much of the literature identifies a need to bridge the "gap" between these worldviews. Another relates to the determinist component of the HGP. According to molecular biologist Walter Gilbert, the HGP answers the question about what actually specifies the human organism, and differentiates human beings from animals. In other words, it allows us to know what makes us human.[5] However, this has raised concerns. "Does this way of understanding the question about what makes us human, and the power it attributes to the HGP in answering this

question, harbour a reductionistic, deterministic view of human life, and of human health and disease, as well?"[6]

Completion of the HGP will have a tremendous impact on every aspect of human health and disease. It will also have a major effect on how medicine deals with disease. One of the unique aspects of the union of molecular genetics and human medicine is the possibility of predicting diseases that will occur sometime in the future. This also, of course, is not entirely new. Genetics has long associated certain diseases with heredity. What is new is the understanding of *why* or *how* diseases are hereditary. What is new is science's ability to identify exactly *where* the root of an inherited disease lies. For example, the root of Huntington disease is a gene lying on chromosome 4. Huntington disease, the first completely dominant human genetic disease to come to light, is also one of the first diseases of genetic origin for which presymptomatic testing became available. "From 1872 to 1993 virtually nothing was known about Huntington's disease except that it was genetic."[7] Since 1993, information about the genetic disease has mushroomed. The dilemma that comes with finding the Huntington gene is that there is no cure. Not a single case of Huntington disease has been cured. Thus, the dilemma: "The acquisition of genetic knowledge is ... outpacing the accumulation of therapeutic power — a condition that poses special difficulties for genetic knowing."[8]

Advances in the HGP will expand the number of presymptomatic tests available, not only for the relatively rare recessive and dominant Mendelian genetic disorders, but also for genetic susceptibility to the more common polygenic and multifactorial diseases, such as cancer, diabetes, atherosclerosis, cardiovascular disease and psychiatric disorders.

However, effective methods to prevent the appearance of these diseases or to treat them when they do appear are often not available. This is one of the biggest challenges in genetic testing for late onset diseases. It opens up a plethora of questions and concerns, most of which this report considers in the next part.

Definitions

Prior to considering the current issues relating to genetic testing for late onset diseases, a few definitions are in order.[9]

A *genetic disease* is a condition that is the result of alterations in the genetic make-up of an individual. These alterations may be the direct consequences of defects in single genes (mutations) or in whole chromosomes, parts of which may be lost, duplicated or misplaced, or of the interaction of multiple genes and external factors.

A *genetic test* aims to detect the presence or absence of, or alteration in, a particular gene, chromosome or a gene product, in relation to a genetic disorder. There are three types of genetic tests:

- *diagnostic genetic testing* of a symptomatic individual to aid in his or her diagnosis, treatment and management;

- *presymptomatic genetic testing*, primarily carried out in healthy or asymptomatic individuals to provide information about that individual's future health, with respect to specific inherited diseases (the purpose of presymptomatic genetic testing is to indicate that an individual has a high likelihood of developing the disease or to exclude it; it is most frequently used in late onset autosomal dominant disorders such as Huntington disease); and
- *susceptibility testing,* which provides information about the genetic component in a multifactorial disorder.

On occasion, test results may be *false positive*, erroneously indicating the presence of a genetic condition, or *false negative*, erroneously indicating the absence of a genetic condition.

Late onset diseases are diseases that normally become symptomatic in adult life.

Single-gene diseases are hereditary diseases caused by a mutation in the allele of a single gene. More than 4,000 such diseases have been described in humans.

Multifactorial diseases are diseases whose genetic components are not the sole cause, but work with other, often environmental, factors in determining a disease outcome. Multifactorial disorders include many cardiovascular diseases, most incidents of Alzheimer disease and some forms of diabetes.

A *carrier* is an individual who is heterozygous (having two different alleles) for a recessive trait. These individuals carry the trait, may pass it on to their offspring, but do not exhibit the trait themselves.

Genetic screening is any kind of test performed for the systematic early detection, exclusion, predisposition or resistance to a genetic disease, or to determine whether a person carries a gene variant that may produce the disease in his or her offspring. Screening programs are distinguished from testing by the approach: health professionals initiate screening, while patients seek help. Screening may be concerned with the general population or with specific subpopulations defined on some basis other than health.

Genetic counselling is a process of consultation by which information is imparted to individuals or families affected by, or at risk of, a genetic disorder. Genetic counselling includes providing information on the nature of the disorder, the size and extent of the genetic risks, the options, including genetic testing, that may help clarify the risks, and the available preventive and therapeutic measures, and providing psychological, social and practical support. In the context of genetic testing, genetic counselling may include responding to the concerns of individuals and their families, discussing the consequences of a test, and enabling individuals to make the optimal decision for themselves, but not determining a particular course of action.

Genetic Testing for Late Onset Diseases

Setting the Context

This section explores 15 issues related to genetic testing for late onset diseases. The literature from which this analysis is drawn covers Europe and North America. The issues were chosen for their constant and consistent presence in the literature. Perhaps the most striking aspect of the literature is the similarity of both the concerns that emerge and the tentative recommendations proposed. The differences appear in the specific details of the recommendations. The goal of this section is to clarify the 15 issues as far as possible within the scope of this project. Each issue would warrant its own project as the issues are full of vast complexities; yet, paradoxically, the essence of each issue is surprisingly simple. This is most likely the reason that there is so much overlap between the European and North American situations.

The International Perspective: Convergence and Divergence

In considering the issues of concern in Europe and North America in relation to genetic testing for late onset diseases, it is striking to note how much convergence exists. While there is a great deal of optimism concerning the potential benefits of genetic testing for late onset diseases, there is a consistent note of caution concerning privacy, confidentiality and the protection of personal data. Concerns relating to discrimination and stigmatization, especially in the context of insurance and employment, are receiving tremendous attention. The continuing growth in the number of genetic tests becoming available and the continual improvement in their accuracy are having a double effect. On the one hand, there is tremendous awareness[10] in Europe and North America of the significant ethical, social, psychological and philosophical challenges that genetic testing for late onset diseases presents. Spurred on by this awareness, a great deal of research is being done by task forces, study groups and expert working groups. On the other hand, there is very little *actual* legislation coming out of these study groups. There is a recognition that "legislation in the field of genetics is hard" to develop because of the "pace of developments and the difficulty of assessing their social consequences."[11] There tends to be continual emphasis on careful scrutiny of what is evolving, yet a "wait-and-see" attitude in terms of legislation. This is reasonable given the experimental state of most genetic testing for late onset diseases and the completely new way of thinking about some issues. However, there are exceptions. The situation concerning insurance is a good example of this trend.

> There is a growing international consensus on the need to restrict the use of genetic information for insurance purposes. Reports and laws differ as to how far these limitations should be implemented. Some European countries prohibit not only the use of genetic testing but also access to genetic information in medical files. Others only prohibit performing genetic tests for insurance purposes (or, more general, outside the medical context).[12]

Thus, generally speaking, the trend in Europe and North America tends toward a basic convergence on the key issues concerning genetic testing for late onset diseases and some divergence in forming guidelines, executing strategic actions and implementing specific policy recommendations.

Current Issues

Public Perception

Public education is necessary to provide an accurate understanding of genetic inheritance and what the notion of being "at risk" means. Confusion about these terms can lead to serious problems such as biological determinism. This is when human development and human behaviour are determined by a person's DNA sequence or one's intolerance of disability. Genetic testing becomes a tool for parents to decide whether to have only "disease-free" and "disability-free" offspring. In order for the public to make informed choices, public education needs to provide information on both the scientific validity of these beliefs and the social consequences of adhering to them.

There are additional, more immediate, reasons to highlight the importance and the need for public awareness of factors involving genetic testing for late onset diseases. There is general confusion about the potential benefits and risks of genetic testing. There is also confusion concerning the *meaning* of genetic testing for late onset diseases. Genetic testing for late onset diseases, while almost unanimously regarded as an important medical tool of analysis, is fraught with interpretive problems. Factors such as risk, probability, sensitivity, specificity and predictive value are difficult to interpret with precision.[13]

One key difficulty is the understanding of predisposition. "Predisposition in the clinical sense is a statistical risk calculation, not a prediction."[14] In 1993, there was an initial report of an increased risk of Alzheimer disease in association with the APOE-ϵ4 allele. In the wake of this discovery, many asymptomatic individuals in the United States and Europe began asking their physicians for the Alzheimer gene test.[15] As well, many people in the United States and Europe wanted to take advantage of the commercial interests that began marketing APOE genotyping. There was clearly a lack of understanding on the part of the public of the many factors that mitigate an individual's risk of future Alzheimer disease.[16]

This being so, it is still certain that the predictive potential of genetic testing for late onset diseases will only increase, although public understanding of the *implications* of this increased predictive potential tends to be limited. Results of a survey conducted to assess attitudes toward genetic testing for colon cancer risk demonstrate a lack of awareness and/or concern for the broader implications of genetic testing for late onset diseases. Many people judge genetic testing as merely another medical procedure without a clear understanding of the potential ramifications in areas such as employment, insurance and relationships.[17]

The Human Genetics Advisory Commission (HGAC) in the United Kingdom also has emphasized the importance of raising public awareness of genetic issues. In the proceedings of its first national conference in 1998, the HGAC emphasized the importance of education "in enabling more groups in society to participate in and follow debates about complex genetic issues." The Commission proposed a participative approach, working in syndicates to ensure "that all groups in society feel they are able to follow and contribute to consideration, debate and discussion of issues."[18] The HGAC highlighted the importance of the education sector in fulfiling this need and is making efforts to collaborate with educators to bring about concrete steps to improve information and public debate. Clearly, public awareness of and education on genetic issues is an important priority in the United Kingdom.

Other European countries have varying degrees of public awareness. Most have formed ethics committees or human genetics societies to promote awareness and understanding of issues concerning human genetics in general and genetic testing in particular. Denmark has a high level of public awareness of issues concerning genetic testing. The Danish Council of Ethics is highly active in facilitating debate-generating activities. The Council conducts large-scale programs to heighten public awareness. The Council of Europe, in its *1992 Recommendation No. R(92)3 of the Committee of Ministers to Member States on Genetic Testing and Screening for Health Care Purposes*, recognized "the need for education of the members of the health care professions and the general public about the importance of genetic factors to health, and for including this subject in curricula for general and further education, both at school and at the university level, and in professional training."[19] The World Health Organization in its *Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services* also highlights the importance of education. "Education about genetics for the public and health care professionals is of paramount importance."[20]

In the United States, the Centers for Disease Control and Prevention (CDC) issued a 1997 report entitled *Translating Advances in Human Genetics into Public Health Action: A Strategic Plan*. Recommendation 4 deals with developing a strategy for communication about genetics, as follows:

> In collaboration with CDC's Office of Communication, conduct a comprehensive review of communication research in genetics, develop a plan for assessing the information needs of various audiences, develop messages, and select media for disseminating information about genetics and public health. Use the Internet as one distribution mechanism. These activities will ensure that the dissemination of information is coordinated, accurate, and timely.[21]

All European and North American countries involved in genetic research and testing for late onset diseases recognize the importance of public understanding and awareness of the political, social and ethical issues involved. The provision of vast amounts of information by councils or working committees is indicative of this need for "clear, unbiased, accurate, and relevant teaching and learning resources, especially interactive resources geared to enabling people to follow complex debates."[22] Some countries are taking steps to make information available at the

public school and university levels. There is a recognition that there needs to be a realistic appreciation on the part of the general public of the balance between the benefits and risks of genetic testing for late onset diseases.

Reasons for Requesting Genetic Testing

People who request genetic testing for a late onset disease perceive themselves to be at risk of developing or passing on a hereditary disease. When individuals perceive this risk, there are several reasons they request genetic testing. One reason is medical. There is the possibility of early treatment or preventative measures for some late onset diseases. For example, early detection of susceptibility to breast cancer gives a woman the option of taking preventative measures. Another reason to request genetic testing for late onset diseases is personal. A person may request testing to relieve the uncertainty of not knowing. Whether the results are positive or negative, some people feel they will be in a better position to make major life decisions. Some people feel that even a positive result is better than the uncertainty of not knowing. Another reason for requesting genetic testing for late onset diseases is to benefit family members either now or in the future. The continued risk and uncertainty of an individual affects that individual's family. Testing not only relieves the individual from uncertainty, but it also alleviates the uncertainty in both current and future family members. As convincing as these reasons are, there are many people who opt not to be tested since they prefer not to know about their risk for late onset diseases.

Many people seek genetic tests for late onset diseases because they want to plan their future. Even a positive result for a genetic disease that is untreatable allows some individuals to set in place a plan of action. People are able to settle conflicts and develop deeper intimacy in their relationships, put their finances in order, settle employment issues and take care of other personal issues.

There is evidence[23] that people who consider themselves at high risk are the most interested in genetic testing. This is due to an immediate sense of susceptibility (for example, a family member is currently suffering from the disease or has recently died.)

There is also evidence[24] that people with higher income are more interested in being tested. There could be some correlation with higher education and the ability to perceive the need for long-range planning, and also the means to put the long-range plan in place.

Assessing the Risks, Benefits and Usefulness of Genetic Testing

Assessing the risks and benefits of genetic testing for late onset diseases and its usefulness is imperative to allow both providers and those being tested to make informed decisions. There are potential benefits for those being tested. For example, family members may be grateful to learn about increased risks, and the information could be very useful to spouses making decisions concerning reproduction or career choice. However, there are potential harms. The information could become available to employers or insurance companies or it could be very damaging psychologically and interfere with family relations. Certainly, there is a general consensus of the benefit of genetic testing for late onset diseases as a critical first step in providing treatment and preventing disease. However, perspectives concerning the *predictive*

value of genetic testing vary. For example, one critic questions the relevance for most women of genetic testing for breast cancer. Testing positive for one of the cancer-linked DNA variants does not mean that a woman will inevitably develop breast cancer. Concomitantly, testing negative does not guarantee that a woman will not develop breast cancer. Indeed, it is suggested that testing positive creates its own dilemmas in terms of procedure.

> "Early detection" is problematic because it is uncertain what is actually being detected, and even such extreme measures as "prophylactic" bilateral mastectomy and oophorectomy provide no assurance that a tumor will not develop in the residual tissue. Given the uncertainty of what being "susceptible" signifies, it is hard to know how to counsel women who are trying to decide whether to be tested for a cancer-associated variant of BRCA1. It is also hard to know how to help women integrate the information they may receive from such a test into the context of their lives.[25]

The phenomenon of false negatives and false positives is also an important consideration. This is not unique to genetic testing for late onset diseases, but perhaps the ramifications are more catastrophic when one is wrongly classified as being "at risk" and treated unnecessarily or when one is wrongly classified as "normal" and not treated.

Tremendous public concern about the safety and effectiveness of genetic tests and their usefulness has elicited a response from many governing bodies in the form of recommendations about reducing risks. In the September 1997 final report of the U.S. Task Force on Genetic Testing, four criteria were strongly recommended to ensure the safety and effectiveness of new genetic tests:

1. The genotypes to be detected by a genetic test must be shown by scientifically valid methods to be associated with the occurrence of a disease. The observations must be independently replicated and subject to peer review.

2. Analytical sensitivity and specificity of a genetic test must be determined before it is made available in clinical practice.

3. Data to establish the clinical validity of genetic tests (clinical sensitivity, specificity, and predictive value) must be collected under investigative protocols. In clinical validation, the study sample must be drawn from a group of subjects representative of the population for whom the test is intended. Formal validation for each intended use of a genetic test is needed.

4. Before a genetic test can be generally accepted in clinical practice, data must be collected to demonstrate the benefits and risks that accrue from both positive and negative results.[26]

Similar concern for the safety and effectiveness of genetic testing for late onset diseases is evidenced in various international and European organizations and in European countries.[27]

Two significant criteria for assessing the usefulness of genetic testing for late onset diseases need to be highlighted. The first relates to the efficacy of disease prevention. Early detection of a future disease may mean that treatment can be used to slow down the progression of the disease or to stop the onset of the disease completely. However, for most late onset diseases, this possibility is rare. The second criteria relates to the right of a person to know his or her own genetic heredity. Knowing the potential risk for developing a disease later in life provides information that a person may feel they need to make decisions about his or her life. The current potential of genetic testing for late onset diseases for satisfying these criteria is minimal. Most late onset diseases are incurable. At this point, there is no known treatment or often very little in the way of preventative measures. Predictability is also tentative. The example quoted above concerning breast cancer is indicative of this. Undoubtedly, science's ability to come up with treatments for incurable diseases and to predict with more certainty will only improve.

One final issue regarding the risks, benefits and usefulness of genetic testing for late onset diseases concerns the availability of genetic information. DNA analysis is increasingly becoming an important resource not only for individuals who want to know their risk for a late onset disease, but also for social institutions in which the individual is involved. This brings to the surface many issues concerning privacy and discrimination — the way in which genetic information is handled and how it is used. The boundary between legitimate use and abuse needs to be continually monitored.[28]

Scientific and Clinical Reliability and Validity

The medical and psychological impact of genetic testing for late onset diseases and the tremendous "normal" variation in DNA warrant increased attention to the importance of scientific and clinical reliability and validity of testing. In addition, one gene for one disease is rare. Even with Huntington disease, which was the first completely dominant human genetic disease to come to light and whose predictability is almost certain, there are large variations in the expression of this genetic mutation. In fact, researchers have found individuals with a mutation at the precise location on Chromosome 4 that has been linked to Huntington disease who do not suffer from the disease. These kinds of discoveries do not undermine the importance of genetic testing for late onset diseases as much as highlight the complexity of the interplay of genes with each other and with social and environmental factors. Thus, caution is warranted against putting too much emphasis on the "predictive" status of test results.

The U.K. Advisory Committee for Genetic Testing emphasizes the difficulty of establishing the validity of testing for late onset diseases because those carrying the genetic abnormality will be healthy for much of their life prior to onset. Regardless, the Committee emphasizes the importance of scientific and clinical validity being clearly established before any genetic test is used in clinical practice, and made several recommendations to this effect:

1. First, because of the high rate of normal variation in DNA, great care must be taken to ascertain that the genetic change found in association with a disorder is causally related and not coincidental. The Committee recommends that validity be based on published, peer-reviewed evidence.

2. Second, even when the scientific validity of the test is clear, there may be other factors that interfere with that validity. The Committee acknowledges that some individuals with genetic change may never develop the disorder, that those with normal test results are still at risk, that it may not be possible to test for all the mutations in a gene, and that some cases may be the result of changes in a different untested gene (what is known as genetic heterogeneity). Consequently, the Committee highlights the importance of knowing "the extent and limitation of the association between the test result and the disorder (false positive and false negative rates)."[29]

3. Third, the difficulty in using linked genetic markers is highlighted by the Committee. The error rate needs to be accurately known and it must be small.

4. Fourth, any information that pertains to the severity of the disease or the specific age of onset is very important to the person being tested. However, the Committee cautions against this forming part of the test result given to the individual unless "the associations have been validated and ... the information can be used in interpreting an individual result rather than an overall series."[30] The Committee mentions two other considerations. Those being tested should be made aware of the technical or human error rate in testing. In addition, when possible, the Committee recommends that "the disorder and particular genetic change being tested for should be confirmed as present in an affected family member."[31] This knowledge facilitates the accuracy of the test result.

The U.S. Task Force on Genetic Testing also emphasizes the importance of safety and effectiveness of new genetic tests. In addition to making recommendations concerning the validity of genetic tests, the Task Force highlights the importance of institutional review boards approving the protocols for the development of genetic tests for late onset diseases. New testing under development should be conducted in certified laboratories.

In Canada, there are also concerns that the implementation of genetic testing only occur after safety and effectiveness are assured. Timothy Caulfield calls attention to fears of Canadian consumers that genetic testing, due to commercial pressures, will be introduced before testing has been scientifically proven to be valid and safe and before ethical and social issues concerning the testing have been thoroughly considered.[32]

Regulation and Monitoring of Laboratories

One key difficulty in regulating laboratories undertaking genetic testing for late onset diseases is that current requirements are not specifically designed for genetic testing. In the United States, laboratories are not required to participate in quality programs for genetic tests, although most do voluntarily. Currently in the United States, "providers and consumers have no assurance that every laboratory performs adequately."[33]

To address this concern, the U.S. Task Force recommends that "no clinical laboratory should offer a genetic test [outside of an investigative protocol] whose clinical validity has not been established."[34] The responsibility falls on the directors of clinical laboratories to ensure the

analytic validity of a genetic test before their laboratories make the test available for use in clinical practice. The Task Force also recommends that before a laboratory routinely offers a genetic test, it should "conduct a pilot phase in which [the laboratory] verifies that all steps in the testing process are operating appropriately."[35] The data from the pilot phase must confirm that the laboratory is competent to perform the test and that the results are in line with those of other laboratories.

The Task Force recommends that genetic testing for late onset diseases be subject to stringent personnel and quality-control requirements. In order to adequately monitor laboratory performance, the Task Force recommends standardization across the United States. Thus, a national (rather than state) accreditation program is required to test proficiency and conduct on-site inspection.

In the United Kingdom, similar concerns about regulating laboratories are in evidence. The U.K. Advisory Committee for Genetic Testing recommends that laboratories be certified, participate in internal and external quality control programs, and be audited to ensure compliance with set standards. The Committee also recommends that laboratories offering genetic testing for late onset diseases be linked to other genetic services and that general laboratories not perform genetic testing. Likewise, research laboratories should not normally perform genetic testing for late onset diseases except when, because of the rarity of the disease, testing is not performed elsewhere. In these cases, research laboratories need to follow the procedures of accredited service laboratories. In addition, the Committee highlights the importance of "equitable and satisfactory" access to genetic testing services throughout the United Kingdom.

In a 1999 report written for Health Canada, Peter Bridge highlights the need for formal training and board certification in genetics for individuals responsible for laboratories that offer genetic tests. In addition, Bridge draws attention to the importance of making genetic testing for late onset diseases more widely available. He highlights the need to develop mechanisms for "interprovincial testing of dispersed families or population sub-groups so that all people have equal access to tests that are deemed suitable."[36] He goes on to say that he has "often been the only person in Canada to offer tests for certain rare disorders."

Direct Marketing

Direct marketing of genetic testing for late onset diseases refers to making tests directly available to the public without the supervision of a trained genetics professional. This raises several concerns. The U.K. Advisory Committee for Genetic Testing recommends against "over-the-counter" genetic testing for late onset diseases. The Committee sees it as inappropriate given the complexity of both the scientific and general issues. The Committee recommends that "any medically qualified person requesting or providing genetic testing for late onset disorders should ensure that they, or another suitably qualified medical practitioner, are actively involved in the process."[37] As a precautionary measure, the Committee wants to ensure the involvement of a trained professional at all times. Another important issue that emerges is the importance of pre- and post-test counselling. The loss of input from trained professionals leads to misunderstandings and undermines the safety and effectiveness of genetic testing.

Another key issue here is the provision of genetic information without proper counselling and follow-up. Test results can cause significant psychological distress. Also, individuals generally do not understand the implications of test results in relation to privacy issues, discrimination and the effect on their family.

The U. S. Task Force on Genetic Testing is also concerned about direct marketing. In the United States, many clinical laboratories advertise the availability of genetic tests for late onset diseases directly to the public.[38] The Task Force cautions that "great care must be taken that information on genetic tests presented directly to the public is accurate and includes risks and limitations, as well as benefits."[39] One key concern of the Task Force is that there is no mechanism in place to monitor the accuracy of the information provided directly to the public.

In the United States, it is also possible for lay people to collect their own specimens and send them directly to the laboratories (although the majority of laboratories do not accept them). The Task Force recommends against this, and discourages advertising or marketing of predictive genetic tests directly to the public.

Another serious consideration is the relative ease with which DNA information can be acquired as a result of direct marketing. It is suggested that the complexity and the potential loss of autonomy that this easy access to DNA information represents signify both a general lack of understanding of the impact of genetic information and how access to DNA information by certain institutions and organizations can be detrimental to the individual.[40]

Genetic Counselling and Consent to Test

Genetic testing for late onset diseases requires support at both the pre- and post-test levels. Initially, an individual being tested needs to understand completely the disorder for which he or she is being tested. *Informed* consent, which is required everywhere in North American and Europe, is only possible when the asymptomatic individual clearly grasps all aspects of the testing — its consequences and limitations and its scientific and clinical validity. In addition, individuals need to be fully informed about the non-medical adverse consequences of testing positive: for example, discrimination, impact on family members and health and life insurance issues.

Both the U.K. Advisory Committee and the U.S. Task Force strongly recommend written informed consent for genetic testing for late onset diseases,[41] both in the context of genetic testing research and testing in clinical practice. Consent should be specific and the individual should only be tested for what he or she has requested. When results from other genetic diseases are also generated from the test, the individual should be informed of this. When an individual is unable to give consent, it is often recommended that the testing be put off until consent can be given. In the U.K., when that is not possible the individual's physician makes the decision on behalf of his or her patient. In Canada and the United States, it is the guardian or closest relative who makes this decision.

Along with the written consent provision, the U.K. Advisory Committee recommends "face to face" meetings with genetic counsellors during which the individual being tested is given complex information. The encounter ensures that the individual being tested has adequately grasped the implications of the test. The Committee also recommends that the individual be given sufficient time to digest the information he or she receives before testing takes place.

Both the U.K. Advisory Committee and the U.S. Task Force place significant emphasis on personal autonomy.[42] In fact, the principle of autonomy in genetic counselling is of paramount importance and is probably the principle most consistently referred to in the context of genetic testing in Europe and North America. Most international organizations also consider it paramount.[43] A person must understand that testing is voluntary. Under no circumstances should a person be coerced or intimidated by a third party (e.g. family or employer or insurance company) into being tested. In addition, a person's decision must be respected and his or her care not jeopardized regardless of the decision.

Post-test support is often required not only for individuals with positive test results but also for those who receive negative results. Those who test positive need help to absorb the results. Individuals need help with the psychological distress they may feel. Also, some people will need to be given important information about measures they can take to alleviate their situation. Those who test negative often need support because they experience "survivor's guilt." This is often so because close family members have tested positive, are currently coping with the disease or have died from it.

There are various means to support individuals who test positive for late onset diseases. Voluntary organizations involved with genetic diseases are an important source of support. Support groups are often available for those who have tested positive and their families. However, perhaps the most significant and effective form of support is genetic counselling. "Genetic counsellors are educated at the master's level in human genetics with an emphasis on the psychosocial aspects of genetic conditions or birth anomalies."[44] There are more than 1,400 professional counsellors in Canada and the United States. In some sense, genetic counselling is both educational and psychotherapeutic. It helps individuals accept and adjust to the knowledge of their predisposition to a late onset disease. Genetic counselling is practised in various ways in Canada, Europe and the United States. Thus, people's perception of the role of genetic counsellors varies from thinking that they merely communicate genetic risk information to thinking that they provide short-term psychotherapy and support. Given future trends in genetic research, various levels of education and assistance will be required for individuals seeking genetic testing for late onset diseases. One genetic counsellor has said the following:

> The future of genetic counseling lies in counseling excellence, professional education, further specialization, anticipatory guidance, and research and academic development. In the new millennium, these emphases will ensure that genetic counseling plays an enhanced role in the future delivery of genetic testing.[45]

Testing of Young Children and Adolescents

Testing of healthy children for late onset diseases warrants points of reference quite different from those raised by testing of sick children. One wonders under what circumstances the testing of a healthy child for a late onset disease would be helpful. Certainly, when early recognition of a disease can lead to some preventative or delaying measures, then it is warranted. However, when there is no useful medical intervention for the late onset disease, predictive testing of a child could result in extremely damaging consequences.

Professional organizations have issued several statements in response to parental requests for genetic testing of healthy children and adolescents. The World Federation of Neurology and the International Huntington's Association reached a consensus decision in 1989 and 1990 not to test children. In 1994, the board of directors of the Clinical Genetics Society of Britain published a statement expressing concern about testing of children. Expressing similar concern were 1995 statements by the boards of directors of the American Society of Human Genetics, the American College of Medical Genetics and the Council on Ethical and Judicial Affairs of the American Medical Association. Authors in the United Kingdom and the United States have recommended that testing be performed only when there is a clear benefit for the minor.[46]

There are several key concerns related to the testing of asymptomatic children for late onset diseases. First, testing children robs them of the opportunity to make their own decision about testing when they are adults. Also, a child has no control over the dissemination of the test results. Another concern involves the problem of discrimination. Even a well-meaning family may treat a child differently once members know of his or her predisposition to the late onset disease. This, in a sense, robs the child of some experience of "normalcy." Or, when a child tests negative, he or she may be (unintentionally) excluded from a family or experience exclusion when a sibling tests positive. Perhaps the most concern arises when children are tested for a late onset disease for which there is no medical treatment. How will this affect both the child and the family?

Despite these concerns, there is evidence that many people, including physicians, consider testing of asymptomatic children to be the parents' decision. In a 1995 "survey of 1,084 U.S. genetics-services providers, 44 percent reported that they had received requests to test children for adult-onset disorders."[47] However, most laboratories in the United States have no comprehensive policy regarding the testing of children for late onset diseases.

Despite professional policy statements cautioning against genetic testing for late onset diseases and recommendations by both the U.K. Advisory Committee on Genetic Testing and the U.S. Task Force on Genetic Testing against testing asymptomatic children for late onset diseases,[48] some consumer groups are in favour of parents' right to test "on the basis that parents are better able to predict the psychosocial outcomes of testing than are physicians."[49] In addition, the majority of primary care physicians in the United States "believe that parents should be able to have their minor children tested for all genetic disorders, including untreatable adult-onset disorders such as Huntington's disease or Alzheimer's disease."[50]

It is interesting, in this context, to note that autonomy is considered so paramount that Canadian genetic professionals feel they much cede to a patient's wishes (or, in the case of minors, to their parents' wishes) even when it means breaking the law.[51]

In Norway, genetic testing of asymptomatic children for late onset diseases is illegal. In contrast, although the U.K. Advisory Committee on Genetic Testing does not recommend the testing of asymptomatic children for late onset diseases, it suggests that professionals who receive requests from parents or persons with parental responsibility should consider the request, but if consent is given to test, the test must be in the child's best interests.

In some cases, the situation with adolescents is significantly different. The Committee recommends that "requests from adolescents deserve full and sensitive discussion, taking into account the individual and their family."[52] Very often an adolescent will be competent to give consent. However, the Committee advises that when there is no clear benefit, the testing should be delayed until the adolescent reaches adulthood.

Prenatal Genetic Testing

The goal of prenatal diagnosis is to have a "normal" child. When prenatal diagnosis delivers a negative result, meaning the fetus is "free" of genetic disorders, prospective parents choose to have the child. When faced with positive results, parents in most Western countries have the option to abort the fetus. Prenatal genetic testing for late onset diseases pushes the "normal" criteria beyond immediate genetic disorders to future disorders. The social and cultural issues surrounding this issue are immense.[53]

Prenatal genetic testing for late onset diseases is relatively uncommon. Often a prospective parent requesting this test does so because of personal or a relative's experience with the late onset disease. This type of request is not the same as the common request to test for serious childhood genetic diseases. The U.K. Advisory Committee for Genetic Testing highlights the importance of only undertaking this form of genetic testing "in the context of full genetic counselling."[54]

The implications of prenatal testing to predict future diseases are significantly different from those of testing children and adolescents. However, one particular aspect has a parallel. When parents request prenatal genetic testing for a late onset disease and opt to continue with the pregnancy despite a positive result, the issue of the child's autonomy is in question. Most governing bodies and professional organizations recommend against genetic testing of children for late onset diseases.

One additional problem concerning prenatal genetic testing for late onset diseases is its potential link to eugenics. David Roy, citing an English specialist in pediatric genetics, discusses the difference between desiring healthy children and reducing the number of defective babies[55] (or, in this case, the number of future defective adults). The goal of prenatal diagnosis of childhood diseases is the former. Yet, what is the relationship between the goal of prenatal diagnosis and that of prenatal genetic testing for late onset diseases? Prenatal genetic testing for late onset diseases provides parents with the option of having a child free of a genetic disorder

that will manifest later in life and will possibly be passed on to future generations. The question here is whether there is a relationship between this and eugenics.[56]

Research Studies

Research studies involving genetic testing for late onset diseases present unique issues that call for special diligence and prudence on the part of researchers and physicians. Consistently, research ethics committees and advisory bodies in Europe and North America recognize the unique character of this type of research and the need for guidance. The special nature of research involving genetic testing in general, and testing for late onset diseases in particular, requires emphasis on three areas: the design of the research, the information required for the participants and consent.[57]

The design of research involving genetic testing needs to include adequate protection to safeguard the anonymity of the research participants and the confidentiality of the research results. Also, research protocols need to be clear about whether test results are to be given to the participant and whether there are any circumstances in which test results would be put in the participant's medical record. The U.K. Advisory Committee advises that, because of the provisional nature of research studies, results should not be given to participants "unless a clear and specific arrangement has been made at the onset."[58] In addition, because of the nature of genetic testing for late onset diseases, an individual's "right not to know" needs to be respected. The U.K. Advisory Committee recommends that research protocols address this issue when results *are* to be disclosed. Due to the stability of DNA specimens, research protocols need to be specific about using stored samples for future tests. The possibility of using samples for multiple genetic testing also needs to be addressed. Due to the hereditary aspect of late onset genetic diseases, involving families in research studies brings up concerns that individuals may be pressured to participate in the research.[59]

The ramifications of informed consent are key in research studies involving genetic testing for late onset diseases. The U.K. Advisory Committee suggests "discussion" rather than merely providing information. It recommends that participants be informed of the individual benefits and potential disadvantages as well as the larger benefits to humanity of this kind of research.

In research studies for late onset diseases, the validity of consent to genetic testing is utterly dependent upon the participant adequately understanding complex information about the research. Written consent provides documented evidence not only of consent but also of the person's grasp of the information. The U.K. Advisory Committee recommends that consent be specific to the disorder being researched, and when research involves a group of allied disorders, this needs to be made explicit. Issues concerning research involving genetic testing of asymptomatic children and adolescents parallel the issues already mentioned under the sections of this paper dealing with genetic testing of children and adolescents for late onset diseases. Always, the decision must hinge on the child's "best interests," which, along with medical interests, also include social and psychological interests.

In the United States, the Task Force on Genetic Testing identifies institutional review boards (IRB) as "the most appropriate organizations to consider whether the scientific merit of protocols for the development of genetic tests warrants the risk to subjects participating in the research."[60] Thus, the U.S. Task Force recommends that protocols for the development of genetic tests for late onset diseases must be approved by IRBs when there is a need to use research subjects and when the test will be made readily available for clinical use. As does the U.K. Advisory Committee, the U.S. Task Force highlights the importance of the human subject in all research studies.[61] Two points of concern need to be considered by the IRB: "the protection of human subjects involved in the study, and the collection of data on analytic and clinical validity, and data on the test's utility for individuals who are tested."[62]

The U.S. Task Force recommends collaboration among testing organizations (for example, the sharing or pooling of data) to expedite data collection. The requirements for IRB review of multicentre collaborative protocols for genetic test development should be streamlined to reduce costs and expedite the studies. Measures should be set in place to facilitate and support collaborative efforts.

Since efforts to ascertain the absolute validity and utility of genetic tests will take a long time, genetic tests will be approved when preliminary results indicate merely the likelihood of validity and utility. Thus, the U. S. Task Force recommends that surveillance of the validity and utility of genetic tests continue even after the tests have been approved for marketing until more definitive results are obtained.

In order for tests to enter clinical practice, test developers need to "submit their validation and clinical utility data to internal as well as independent external review bodies." As well, "test developers should provide information to professional organizations in order to permit informed decisions about routine use."[63] The Task Force suggests reviews at both the local and national levels.

Population Screening

With expanded capacity to identify genetic changes, testing for late onset diseases is no longer confined to those at risk in a family context. Genetic screening for late onset diseases is becoming increasingly possible for a large number of disorders. Screening is generally initiated by public health authorities and looks at the general population or specific subpopulations in relation to certain health concerns. As with genetic testing for late onset diseases, the benefits of presymptomatic genetic screening are early detection, possible preventative or delaying treatment, and being able to make informed reproductive decisions. The detrimental effects are anxiety and other negative psychological reactions mostly due to predisposition to a disease that is not treatable.

Three principles have been suggested to guide public health decisions regarding genetic screening.[64] First, genetic screening should ultimately improve the health of the population. Genetic screening is practised in terms of populations, not individuals; therefore, the health of the whole population is the major concern, not that of particular individuals. This underlying principle affects precisely what, if any, late onset diseases call for genetic screening. The second

principle deals with efficient and just use of resources. Again, with the population in mind, not individuals, the principle of equitable use of scarce resources is highlighted. Currently, most late onset diseases are exceedingly rare. Thus, the investment of resources in the population as a whole is questionable. However, the growing discovery of a genetic base for more common and costly diseases, for example heart disease, will dramatically change this. Third, the target population needs to see that the benefits of genetic screening outweigh the costs. Since most late onset diseases are currently untreatable, the benefits are ambiguous. Yet, the genetic base for common diseases may, again, quickly change this scenario.

As the technology behind genetic testing simplifies, screening large groups and even whole populations for late onset diseases will become more prevalent. The U.K. Advisory Committee on Genetic Testing suggests that there are special concerns that this prospect raises: for example, "greater chance of error in sample collection and laboratory analysis" and large numbers inhibiting the "particular need to ensure full understanding and support." Also, there are ramifications for families of those being tested, especially when the individual being tested is unaware of the genetic component of the disorder. The Committee recommends that genetic screening for late onset diseases only be introduced when "an appropriate and effective treatment (of follow-up action) is available, and where treatment of the disorder at an early stage is of more benefit than treatment at a later stage." The committee also recommends that support be available for individuals who test positive. Also, understanding the significant difference between health care and public health, the Committee highlights the potential tension between individual autonomy and public health concerns.

The idea of population screening raises concerns about "the relative ease with which information on DNA sequences can be acquired, when adequate knowledge of its correct interpretation is lacking."[65] One current example is the prospective population screening for BRCA1 and 2 gene mutations. In light of still incomplete understanding of the implications of a positive or negative test result, "genetic testing and population screening are not recommended."[66] However, testing is quickly becoming commercially available in Europe and North America, and is being "widely used before safeguards are in place and the clinical implications are fully understood."[67]

Confidentiality

Privacy is contingent on social relations, and is affected by many factors.[68] However, technological innovation has rapidly focussed our attention on the importance of privacy. "Technological advances in fixing image and sound and in intercepting and recording speech (notably phone conversations), as well as the progress of statistical methods, have enhanced the capacity of intrusion in the private life of individuals."[69] Intrusion into the private lives of individuals has grown exponentially with the increasing power of molecular genetics and biotechnology.[70] The ramifications concerning privacy and confidentiality are tremendous, and significant aspects of these ramifications are directly related to the unique situation of asymptomatic persons testing positive for late onset diseases.

There are several key concerns related to genetic testing for late onset diseases and the issue of privacy. Perhaps one of the unique problems in this area pertains to the "intergenerational" nature of genetics[71] and subsequent dilemmas concerning invasions of the privacy of those who do not choose to be tested. Another unique dilemma concerns the lack of clarity of boundaries between medical and non-medical information when identifying individuals or groups of people predisposed to late onset diseases. This information is of interest to many "third party" individuals or organizations. Third-party interest tends to be motivated by three things: implications for family members, discrimination and financial interest. One final area of concern is the "particular threat to privacy and confidentiality ... when genomic information on individuals is, or will be, stored in computerized genetic registers."[72]

The issue of confidentiality is crucial. In recognition of the tremendous importance of confidentiality in all aspects of genetic testing, there is a vast amount of study being done as governing bodies ponder the need for special legislation.[73] The U.S. Task Force on Genetic Testing recommends that test results "be released only to those individuals for whom the test recipient has given consent for information release."[74] The Task Force stipulates that care should be taken to ensure unauthorized persons or organizations do not gain access to this information. "Under no circumstances should results with identifiers be provided to any outside parties, including employers, insurers, or government agencies, without the test recipient's written consent."[75] This also pertains to the test recipient's family members, except under extreme circumstances. This strong statement reflects the general consensus in Europe and Canada.[76]

There are three interrelated topics that need to be discussed in relation to these concerns: discrimination, employment and insurance. The interrelationship between the issue of confidentiality and the burgeoning problems currently surrounding genetic testing for late onset diseases in relation to discrimination, employment and insurance presents particularly complex problems.

Discrimination

In a Canadian government interdepartmental report entitled *Biotechnology, Ethics and Government*, the authors refer to the creation of a "new social category of the not-yet-ill."[77] The category is being created by the rapidly growing capacity of molecular genetic research to test for late onset diseases. Regardless of the tentative nature of the test results, the creation of this new social category is already leading to problems of discrimination. This is especially seen in the workplace and in relation to insurance. Both are discussed in the following sections of this report. However, discrimination — the practice or an instance of discriminating against people on the grounds of race, colour, sex, social status or age — often has roots in fear of difference, or fear of the other or the stranger. Thus, genetic discrimination — "discrimination directed against an individual or family, solely because of their apparent or perceived variation from the 'normal' human genotype"[78] — potentially denies an individual the opportunity for social benefits such as education, work, life insurance and, sometimes, depending on where the affected person resides, health insurance. However, over and above discrimination in terms of deprivation of social

benefits because a person will eventually not be "useful" or will become a burden according to the standards of a society, the stigmatization of discrimination touches the person him or herself. Fear eventually becomes directed at the person him or herself. This is the root of discrimination — fear of difference. As one author expresses it, "Predictive genetic typing may create an underclass of individuals whose genes seem to have marked them for the nowhere track."[79]

The genetic link within families can also occur in whole communities or whole races. "Genetic diseases are often over-represented in racial and ethnic groups or even in specific local communities."[80] Thus, a predisposition to a genetic disease can exacerbate discrimination that already exists because of a person's racial or ethnic origin.[81] Also, fear of discrimination can play a significant role in an individual's or group's willingness to participate in research or screening programs.

Many governments are attempting to address this problem. The U. S. Task Force on Genetic Testing cautions against "unfair discrimination" that may result from genetic testing, pointing specifically to discrimination that may result because a person has had a genetic test or has received an abnormal genetic test result.[82] The Privacy Commissioner of Canada recommends that "personal genetic information collected by government institutions or private sector physicians providing ordinary medical care should be used only to inform a person's own decisions about medical care. This information must not be used for any other purpose."[83]

The *Americans With Disabilities Act*, implemented in January 1992, limits pre-employment testing to the assessment of a person's actual ability to perform a job. The Act will help to limit the abuse of tests that indicate genetic predisposition in a person with no symptoms. However, control of discrimination requires more than legislation. "So long as persons continue to be conceptualized as aggregates of physical attributes and as gene-transmitting agents, biology can be used as both a standard for opportunity and a justification for discrimination."[84]

One final important point. Individuals with hereditary diseases and disabilities are questioning the implications of discrimination against inherited diseases in terms of their own value. Strangely, in a (Western) culture in which people who are physically and mentally challenged are gaining more and more inroads into society, the trend of discrimination against individuals who may develop a late onset diseases is ominous. "People with hereditary diseases and disabilities fear that increasing access to genetic information through prenatal screening, together with the increasing acceptability of selective abortion of "defective" fetuses, will devalue them and their experiences, leading to increased discrimination against those who are physically different."[85]

Employment

A statement from the Human Genetics Advisory Commission paper *Human Genetics Advisory Commission Second Annual Report 1999* gives witness to the monumental importance of the relationship between genetic testing for late onset diseases and employment. "The implications of genetic testing for employment are the Commission's current priority."[86] Employers requesting health information from prospective employees is not unique. Since the beginning of the 20th century, employers have requested information about potential employees' health. In the United States, both the volume and the sensitivity of employer-maintained employee medical information have grown tremendously. "By the early 1980s, nearly 90% of companies with more than 500 employees were performing preemployment medical examinations."[87] As genetic testing for late onset diseases becomes more extensive and precise, it will provide employers with a powerful tool with which to evaluate current and potential employees. "As a species of health care information, genetic information is particularly sensitive because genetic screening and monitoring reveal much more personal information about the individual than other types of medical surveillance used by employers."[88]

Although there are situations in which genetic testing may be appropriate — that is, when it detects a condition that would put the employee or others in the workplace at risk — the key problem is the potential for unfair discrimination. This is especially a problem for healthy individuals with a predisposition for a late onset disease. The employee or potential employee's ability to remain productive is critically important to employers. The amount of sick leave an employee takes and the cost of (extended) health and life insurance are important factors to employers. The advantage that genetic testing gives employers is significant because it heightens their capacity to judge the potential contribution an individual will make.

Because there are situations in which genetic testing may be appropriate in the workplace, the U.K. Advisory Committee on Genetic Testing resists a total ban. Because completely banning genetic test results for employment purposes "would ... not be in anyone's best interests," the Committee suggests four policy principles be set in place to "provide appropriate protection to the public in a manner which is least burdensome to employers."[89] The principles are that an individual should not be required to take a genetic test for employment and should only be required to disclose previously taken genetic tests when there is clear evidence that the information is needed for the job. Employers should offer a genetic test when there is any potential danger to either the employee or others in the work environment and there must be some way to assure the validity of the test.

The varying definitions of *genetic testing* and *genetic information* currently result in a wide diversity of responses from jurisdictions within the European Union. In Austria, employers and insurance companies are prohibited by law from collecting, demanding or using data derived from genetic tests. In Denmark, legislation aims to ensure that health checks focus on actual or current health conditions, and that those conditions are relevant to the employee's work. In Finland, recommendations have been tabled that employers should not be allowed to subject job seekers to genetic testing during recruitment, or to test employees already hired. In France, bioethics legislation specifically prohibits access by any third party, notably employers and insurance companies, to information held in data banks, and makes it illegal for them to ask

individuals to provide such information. In the Netherlands, the *Medical Examination Act* of 1997 prohibits employers from applying medical criteria to recruitment unless there is an unambiguous health requirement for the job. In Norway, genetic testing in the workplace is illegal. In Spain, legislation makes provisions to distinguish between predictive testing for general health and testing for the protection of workers who are especially sensitive to specific work environments.

There is also a diversity of responses in the United States and Canada. In the United States, 12 states have enacted legislation to prohibit discrimination in employment on the basis of genetic testing or genetic information. However, this has had no practical effect.[90] There are currently no federal laws dealing with gathering or using genetic information in the workplace. However, there are a variety of indirect laws that affect genetic testing. In particular, the *Americans With Disabilities Act* has been cited as useful in its dealing with issues of employee discrimination.[91] This is similar to the situation in Canada, where provincial and federal laws indirectly deal with this issue.

Insurance

The use of genetic information for insurance purposes is equally problematic. Insurance traditionally differentiates people according to their risk, and increased genetic prediction in the context of insurance is "likely to grow massively in the future."[92] One report puts it this way: "Insurance is about attributing a monetary value to risk and about reducing risk within reasonable limits. Genetics has, until now, been almost exclusively about predicting and assessing risk. Both seem to be made for each other."[93] The ramifications of genetic testing for late onset diseases affect both Europe and North American insurance practices but to varying degrees. In the United States, the market for private health insurance is, of course, much greater than in Canada or in Europe. For life insurance, however, the situations are similar. Most countries acknowledge the need for some special regulation concerning genetic information. For example, the British government accepted recommendations made in the Human Genetic Advisory Commission report *The Implications of Genetic Testing for Insurance.* The Commission did not suggest "a permanent ban on the use of genetic test results in insurance" but rather that "safeguards were required to ensure that the results of genetic tests could only be used by insurers when a quantifiable association between a given pattern of test results and events actuarially relevant for a specific insurance product had been established."[94] Similarly, the American National Institutes of Health/Department of Energy Task Force on Genetic Information and Insurance does not see a separate status for genetic information succeeding because other varieties of health-related information are equally sensitive. Also, genetic information is not segregated from other health information and, thus, is too easily accessible.

> Policies intended to protect genetic privacy will need to address the privacy of health related information in general. If we want strict standards to safeguard genetic information, then those same standards will have to extend to all health related information.[95]

Belgium and Norway have gone beyond mere safeguards. They have opted to prohibit insurance companies from requiring genetic testing or from having access to genetic information. There is no ban in Canada. Generally, insurance companies do not request genetic testing from applicants both because of the cost and the uncertainty of the predictive value.

> Research results, on their own merit, should have little interest for insurers, employers, and others, because without associated data on the consequences of mutations in individuals who are asymptomatic at the time of testing, the results will lack predictive value. For that reason, insurance companies have adopted a wait-and-see approach.[96]

The Netherlands takes the middle road in prohibiting insurers from demanding disclosure of genetic information when the requested insurance coverage is below a certain limit. But for greater coverage, "insurers should be free to demand disclosure of existing genetic information but not to demand genetic testing. The limit can be set appropriately to the applicant's social and financial circumstances or it can be a uniform one."[97]

There are several key issues in the context of genetic testing for late onset diseases and insurance. As noted above, issues of privacy and confidentiality enter into the debate about whether insurance companies should have access to results of genetic testing. Also, at least for now, the reliability of genetic tests is far from absolute. The problem of false positives and false negatives has already been noted, as have the limits of genetic testing in pinpointing the age of onset or the severity of the disease. Thus, the predictive value of genetic information (in terms of an individual's future medical costs) is, at least for now, less valuable to insurance companies than more traditional predictive factors.[98] As with employers' access to genetic information, the problem of genetic discrimination is a key factor in the accessibility of genetic information to insurance companies. In addition, when an individual seeking insurance is required to disclose genetic information, that person may choose not to seek out genetic testing or genetic counselling. Another problem is establishing safeguards to ensure that insurance companies keep genetic information confidential.

Perhaps the most important question concerning insurance companies and genetic testing is whether genetic information should be treated differently from other medical information. It has been suggested that what differentiates genetic information from other medical information is its predictive power. For example, the European Parliament, the Royal Norwegian Ministry of Health and Social Affairs and the Council of Europe assert that insurance companies should not have access to genetic test results nor should they have the right to require genetic tests. These legislative bodies have not taken the same approach to other types of medical data because of the wide scope of information genetic testing can provide. It is argued that there is a combination of factors responsible for the difference between information obtained from standard medical tests and from genetic tests. For example, genetic testing requires only a small sample, which can be used for multiple tests and be reused indefinitely. As well, "genetic tests are independent of the patient's age and/or clinical state."[99] Therefore, most governing and advising bodies concur that giving genetic information special status, apart from other medical information is necessary.

Insurance companies face a serious problem of people strategically using genetic information concerning future health problems in their applications for insurance policies. The possibility of predicting late onset diseases will eventually modify the consumer's habits. The consumer will benefit from the knowledge obtained through genetic screening by buying larger insurance plans than he or she normally would in order to provide for the future. The insurance companies will be disadvantaged since they do not have access to the genetic information and they will be selling insurance for a lower premium than they normally would if they had access to the medical information. The long-term results of this could undermine the insurance industry and create an economic imbalance.[100] Certainly, the insurance industry is worried. In response to the Alzheimer's Association recommendation that genetic testing be anonymous, insurance companies viewed "these pronouncements with mounting frustration ... calling it a frontal assault on a fundamental business practice."[101] This, of course, affects the insurers as well. "No one would want to buy insurance from an insurance scheme with policies of such a kind that its short and/or long term sustainability is questioned."[102] All are implicated in a potential decline of the insurance industry.

Factors Shaping Policy Recommendation and Implementation

The goal of this final section of the report is to identify factors that are shaping policy recommendations and implementation in North America and Europe. The factors are fairly straightforward and consistent across international boundaries although expressed in a variety of complex manners depending on the particular context. The factors often overlap and at times are in conflict with each other. In order to identify these factors and explore their significance, two tools of analysis are used: technology assessment and a scale of values. In the following two sections, each tool is delineated to show how it sheds light on factors shaping policy recommendations for genetic testing for late onset diseases.

Technology Assessment

Technology assessment is one attempt to control technology through public policy. Public policy refers to what governments decide to do or not to do. "Public policy is a purposive course or pattern of action or inaction by government officials."[103] In order to make recommendations, technology assessment looks at "all the possible and probable effects on society of introducing or expanding particular technologies."[104] At times, the task of technology assessment is reduced to providing an "objectively" sound basis to help governments make decisions about particular technologies. The underlying presupposition is that any particular technology can be judged by considering the costs and benefits. Technology assessment provides a very concrete, accessible tool to analyze particular technologies. In its basic form, technology assessment is concerned with safety and efficacy and whether the benefits outweigh the costs.

However, technology assessment has been criticized for its narrow frame of reference and, in particular, for the limits of the almost exclusively *instrumental* character of current technology-assessment thinking. Therefore, it is important to note that technology assessment is useful in identifying the factors shaping policy development for genetic testing of late onset diseases, but it is merely a first step. Although others have taken more liberty with technology assessment,[105] this paper restricts its analysis to this very utilitarian interpretation. (The scale of values involves a much deeper level of analysis.) Thus, technology assessment is a crucial first step. Often it is this first level of analysis that captures the public attention in a dramatic manner and thus has a tremendous impact on public policy. What are the possible and probable effects on society of genetic testing for late onset diseases? How are these effects influencing policy decisions and policy recommendations?

The purpose of genetic testing for late onset diseases is to identify in healthy (asymptomatic) individuals the likelihood that they will develop a disease later in life. Testing for late onset diseases can take place at any time during a person's life, even prior to birth. In terms of safety, there is virtually no risk in the actual testing. The results present some risk due to the phenomenon of false positives and false negatives. The risk here involves unnecessary distress and unnecessary treatment in the case of false positives, and a mistaken sense of security involving, perhaps, high-risk behaviour in the case of false negatives.

In terms of efficaciousness, genetic testing for late onset diseases is problematic. This is largely, although not completely, due to commercialization of genetic testing. The U.S. Task Force on Genetic Testing is "concerned that genetic tests intended to predict risk of future disease in apparently healthy people were becoming available before adequate data on sensitivity and PPV [positive predictive value] had been collected."[106] Thus, the Task Force established criteria to ensure clinical validity of genetic tests, and recommended that protocols for genetic tests be approved by institutional review boards.

Another factor that influences the efficaciousness of genetic testing for late onset diseases is that genetic tests are rarely 100 percent accurate. The false positive and false negative phenomenon has already been noted. In addition, usefulness is lessened significantly due to the very real possibility of an incorrect interpretation of knowledge that is coming at a rate too fast to integrate.

> Serious difficulties arise from the relative ease with which information on DNA sequences can be acquired, when adequate knowledge of its correct interpretation is lacking. This can be seen in relation to the so-called breast-cancer genes BRCA1 and BRCA2. These two DNA sequences have both been linked to increased susceptibility to breast or ovarian cancer. To date, more than 100 variants of BRCA1 and several variants of BRCA2 have been identified. Only a few of them, however, have been shown to be associated with tumor growth .... We must therefore ask how the predictive tests now being developed on the basis of variants of BRCA1 are relevant to most women.[107]

Similar concerns are expressed about Alzheimer disease,[108] other types of cancer and heart disease.[109] Most governments and advisory bodies are very aware of these difficulties and the risk factors involved in genetic testing for late onset diseases. There is, however, a tension between the commercial efforts to market genetic tests for late onset diseases to the public as quickly as possible and the organizations and individuals who are advising caution. Several policy recommendations are emerging from this situation.[110]

The key benefit of genetic testing for late onset diseases is of course the settling of uncertainty. Whether one tests positive or negative, uncertainty and the anxiety that the result produces is lessened. Certainly, testing is beneficial and legitimate insofar as there are possible treatments to either delay onset or to prevent onset altogether. Also, testing helps individuals make reproductive decisions. Although the number of individuals who develop late onset diseases is a small percentage of the total population, "in high economy societies they account for a considerable proportion of mortality and serious ill health in middle life, often affecting those with major employment and family responsibilities."[111] When the disease is treatable or at least delayed, the cost of genetic testing is justified. However, a large number of late onset diseases are at present not treatable. Is genetic testing for late onset diseases, in the absence of any known treatment, still legitimate in these cases?

Financial considerations concerning genetic testing for late onset diseases are important. Testing may be quite expensive, especially for the needed counselling and follow-up. However, "the cost of genetic testing and prophylactic intervention is trivial compared to the cost of treating these diseases after the fact."[112] The cost of health care and the scarcity of resources are major factors in how genetic testing for late onset diseases is being dealt with at a public policy level. How this will play out in the years to come will depend on the efficacy of the vast number of genetic tests that are currently being researched.

Scale of Values

Values determine the social good or order that is intricately tied up in communal life. Although the term *value* may seem an abstract notion, it is, in fact, imminently concrete. This is because the question of value always emerges when we are faced with concrete decisions. Value is what we are wondering about when we ask ourselves, "Is this good?" or "Is this the right thing to do?" or "How should Canada deal with the rapid development and availability of genetic tests for late onset diseases?" One author describes values as "ideals, experiences or states of affairs pursued by individuals, organisations and communities."[113] Here, the multidimensional character of values is highlighted. Values occur on various levels. Human beings respond to values using a scale of preference. Organizations and communities are aggregates of human beings responding in concert to concrete situations using a scale of preference. Values are ordered hierarchically in the sense that they move from immediate self-satisfaction toward self-transcendence, which is the condition necessary for relationships. The five types of values are as follows.

1. Vital values are the most basic or fundamental values — life itself. It is at this level that individuals are concerned with survival. Values at this level respond to the impetus to stay alive or to stay healthy.

2. Social values are no longer concerned with mere survival. Rather, these values emerge from the desire to order our world. Thus, social values shape the social, political and economic orders and the social infrastructure. They are concerned not with the survival of individuals *per se*, but with the survival of a whole society.

3. Cultural values go beyond social values in that they are concerned not with survival or the ordering of our living together, but with the *meaning* of our lives. Our lives are informed by meaning. Cultural values are the backdrop through which these meanings are discovered, expressed, validated, criticized, corrected, developed and improved.

4. Personal values focus on the individual and are the originating values within a community. Values at this level have to do with personal integrity: privileges, attentiveness, intelligence, reasonableness and responsibility. Personal values refer to the capacity for individuals to go beyond themselves and to affirm something about their world. The endeavour of ethical reflection on an issue such as genetic testing for late onset diseases emerges precisely from our capacity to experience the world and say something about it. We are not isolated monads. We are beings engaged in the

world, and it is precisely personal values that facilitate this engagement. Personal values allow us to realize values in ourselves and promote their realization in others.

5. Ultimate values are implicit in all human questioning, although they are often not articulated. When one is faced with limit situations such as illness and future health, ultimate values are articulated for *ultimate* meaning and value. They are evidenced in the human propensity to grasp meaning beyond ordinary daily existence, particularly at times of limit situations.[114] Ultimate values also push one to consider issues against wider horizons of historical progress and decline.

Perhaps, health is the basic factor shaping policy development in genetic testing for late onset disease. The importance of testing for late onset disease directly stems from the basic, vital value of human existence — health. The immediate goal of genetic testing for late onset disease is to identify whether an individual will develop, late in life, a life-threatening illness. Yet, underlying that goal, is the promotion of health. The intense concern that most Canadians feel about the current crisis in the Canadian health care system stems from this basic and vital value of health. Thus, human health is a crucial factor that directly impacts policy development.

Social values that influence policy development are more complex. The health care system plays a fundamental role in maintaining the social order of a society. Social order maintains the health and well-being of citizens, and the threat of illness undermines that social order. Alzheimer disease is a good example: more than 200,000 Canadians over the age of 65 are afflicted with that disease.[115] Late onset diseases such as Alzheimer's, in an aging population, threaten the social order of a society. In effect, the tremendous interest in genetic testing for late onset disease stems directly from this very and basic value.

If health care is directly linked to maintaining the social order in relation to the well-being of citizens, the infrastructure of a social order depends just as much on the *economic* health of a society. A U.S.-British declaration on March 14, 2000, stated that the information from the Human Genome Project would be freely available for research elsewhere. This announcement resulted in a significant drop in biotechnology stocks. According to *Philadelphia Inquirer* commentators, what appeared to be an altruistic gesture on the part of the U.S. and British governments had more to do with promoting cooperation between the public and private efforts in the Human Genome Project than with making information available to all. The National Institutes of Health owns dozens of patents on human genes and gene sequences. Thus, the economic well-being of the social order, like the health of citizens, is another factor that shapes policy on genetic testing for late onset diseases. Meanwhile, the concerns of the safety and reliability of the genetic tests are not withstanding, and direct marketing of genetic tests for late onset diseases is already happening in North America and Europe. Those companies that drive this trend have a vested interest in maintaining the social order.

Cultural values incorporate complex meanings that shape and direct lives. They remind us of the sheer plurality of human existence. It is at this level that *human* factors shape policy on genetic testing for late onset disease, and tremendous conflict between various values occurs. The two values that North Americans and Western Europeans covet are autonomy and self-determination — thus the importance of privacy,[116] confidentiality[117] and informed consent[118] for genetic testing. Almost without exception, Canada, the United States and the European countries emphasize the importance of confidentiality and informed consent. These two factors strongly influence policy. In some countries, legislation regarding who has access to genetic information has already been formed.[119] The importance of *informed* consent occurs in every international and national study and in recommendations for genetic counselling.[120] Autonomy and self-determination are cultural values that provide meaning of life for an individual. They also shape our worldview, and we measure what is acceptable by these values.

Non-discrimination is important for policy making, and a tolerance of otherness is highly valued in Europe, Canada and, perhaps slightly less so, in the United States. This is particularly so, as has been shown, in relation to employment and insurance issues. An equally important concern is the stigmatization of individuals who test positive for late onset diseases. The categorizing of the "not-yet-ill" or at-risk individuals is turning attention to cultural understanding of "normality" and "abnormality."[121] While there is consistent sensitivity in North America and Europe to combat stigmatization, there is also a drift towards "geneticization." Geneticization is the process in which differences between individuals are reduced to their DNA codes because most disorders, behaviours and physiological variations are defined, at least in part, by their genetic origin. Geneticization threatens the state of difference among members of the population, which is acutely experienced by individuals who are mentally and physically challenged.

In early 1995, a public debate was held in the Netherlands, which brought together experts and laypersons, including representatives of parent, patient groups and organizations of physically challenged people. The resulting final declaration emphasized the importance of free choice and non-directiveness in counselling, and addressed the concern of uninsurability of those tested with late onset diseases. "The declaration also pointed out the importance of psychosocial support, public and professional education in genetics and ... that cost-containment in health care should not lead to social pressure for selective abortion."[122] In a separate meeting of "patients," parents and physically challenged people in January 1995, "great fear was … expressed by handicapped people that genetic screening would reinforce negative views towards disease and handicapped life, and would lead to more stigmatisation and discrimination." There was concern that this would prevent their integration into society. Hoedmaekers discusses the possibility for limiting "the use of pre-natal diagnostic technologies to prevent negative consequences for handicapped people … and concluded that the freedom of the individual to make moral choices should not be given up…."[123]

In the Netherlands, there is tremendous concern about the protection of individuals' privacy. (This is reflected consistently in all policy recommendations in Europe and North America.) Recommendations from the previously mentioned public debates in the Netherlands highlight this concern. They also promote a social climate that favours acceptance and respect of physically challenged people.

The situations with discrimination and stigmatization in the Netherlands reflect the tension that is common in North America and Europe during policy making for genetic testing for late onset diseases. In North America and Europe, there is a gradual tendency to manage health care through the intervention of genetic technology. The concerns and policy recommendations from the study groups and task forces indicate that although this tension surfaces consistently, it is not recognized.[124]

The tensions between conflicting values highlight the limitations of cultural values. These tensions cannot be resolved at the level of cultural values, but only at a higher level on the scale of values. Although it is beyond the scope of this report to attempt to resolve these tensions, this topic needs to be addressed in order for one to understand and assess various factors that emerge from genetic testing for late onset diseases. These factors would ultimately shape policy recommendations and, in some cases, lead to legislation in North America and Europe.

One extremely useful tool to enhance awareness of tensions between conflicting values is to differentiate the values along a scale of preference. Vital and social values are important but not essential humanizing values that create human communities. Cultural values are significantly humanizing. However, they call individuals to a heightened level of integrity to develop creative solutions to problems arising from people's conflicting concerns and agendas. Genetic testing for late diseases presents fundamental concerns about privacy, confidentiality, discrimination and informed consent.

The National Consultative Ethics Committee for the Life and Health Sciences (CCNE), issued a report in 1997 entitled *Genetics and Medicine: From Prediction to Prevention*. The report highlights a shift in levels from cultural values to personal and ultimate values. A long quote from this document provides a sense of the overall direction of the concerns:

> The very tumultuous history of genetics is because its very object is at the heart of the fundamental interrogation which constitutes the human being: where do I come from, who am I, what shall I bequest to my offspring, in what way am I both similar to and different from other people. This is why the science of genetics has had and still has more individual, political, and social repercussions than any other. At the present time, progress in human genetics shows promise of a not very distant future when all the human genes — of which there are approximately 100,000 — will have been identified, located on the chromosomes, and when their functions, or at least their implications in genetic diseases will be almost fully understood. The myth of the gene as the stuff of which life itself is programmed, is such that of it is born the illusion that perfect knowledge of the genome of an

> individual will lead to an understanding of the reality and fate of that individual. Metaphors such as the book of life which would give access to the essential human being if only one could decipher the genetic alphabet and syntax, refer to that notion. Such a concept is scientifically unacceptable and ethically dangerous.[125]

The report addresses the tremendous impact of genetics on society. It links this impact to the goal of genetic testing for late onset diseases, to forecast the appearance of certain diseases before their symptoms are expressed. "However, there are grave uncertainties about the value of the predictions and whether it is truly possible to prevent the conditions, and also whether this form of prevention is truly beneficial to individuals and to society."[126] Certainly the *a priori* benefit of avoiding or delaying the onset of disease is unquestioned. However, the CCNE report analyzes this issue and raises the question of "destiny and freedom in the face of knowledge of genetic risks."[127] The report also highlights the tension between the importance of knowing about the disease (especially when there is something that can be done to slow its onset or perhaps even treat it) and knowing the certainty of an incurable disease's onset. Despite the advantages of knowing what will come and the ability to plan the future with some certainty, the repercussions at the deeper, existential level are momentous.

> The significance of the exercise of freedom by a person whose genetic predisposition leaves no choice but a life in the grip of terrible constraints or preventive mutilation or risk of incurable disease, is open to question. Another individual dimension of a genetic fate revealed is that sometimes, in the case of the handing down of a serious disease, it is tantamount to a curse put on the lineage, since the parents may be considered guilty of transmitting a faulty gene to their children who, in turn, feel guilt at the possibility of transmitting it to their own descendants.[128]

In response to the concerns mentioned above, the CCNE provided the following recommendations: respect individual rights to not know about late onset diseases; understand the effects and impact of being tested for late onset diseases; abstain from testing for non-medical reasons; ensure provision of support such as counselling to alleviate possible psychological repercussions; ensure medical confidentiality including members of one's family; take precaution on data storage; prohibit non-medical uses of the test results; pay attention to how the media informs the public, which may lead to false hopes, and be cautious of commercial interests for genetic information, which can detriment the truthfulness and independence of the genetic information.[129] The final recommendation is particularly relevant in its link to the ultimate concerns of the CCNE:

> Genetic tests give information on the identity of persons and emphasize their diversity which contributes to the rich nature of humankind. To use such information for the purpose of selection or of discrimination in social or economic terms, be that in the realm of public health policies, employment, or insurance systems, would be crossing a boundary of the most extreme gravity and would question those principles of equality of rights, dignity and

> solidarity for all human beings upon which society as we know it is based. The CCNE insists on the necessity of observing those fundamental principles whatever aims may be pursued by genetic testing. Human Rights are at stake.[130]

The protection of human rights and human dignity is the basis for making ethical and social decisions on genetic testing for late onset diseases in Europe and North America. Whether directly or indirectly, the Universal Declaration of Human Rights is consistently being referred to in international and national policy statements on genetic technology and genetic testing.[131]

Conclusion

The “not-yet-ill” is a rather peculiar expression used to describe a new social category of individuals. The justification for this new category is based on a genetic test that identifies a mutation in a gene and thus predicts that these individuals will develop a certain hereditary disease sometime in the future. In exploring the topic of genetic testing for late onset diseases, a multitude of issues come to the fore. The goal of this report has been to elaborate on these issues. Thus, the report has considered the topic from three perspectives. First, it looked at genetic testing for late onset diseases within the larger domain of genetic technology. Second, it considered the key issues that genetic testing for late onset diseases raises. Third, it explored the underlying factors shaping policy recommendations and implementations. Needless to say, given the complexity of every aspect of this tremendously volatile topic, the work has just begun.

Annotated Bibliography

The bibliography is divided into three sections as follows:

1. **Section one** indicates literature that provides *tools for assessing* genetic testing for late onset diseases from various perspectives (for example, ethical and social). It assists in setting up a framework to analyze the complexity of genetic testing for late onset diseases, in identifying the underlying factors that shape policy formation and policy-related issues, and in assessing genetic testing for late onset diseases by providing a long-range perspective.

2. **Section two** indicates literature dealing with the more *general category of genetic technology*. Genetic testing for late onset diseases falls under the broader category of genetic technology. Literature in this category provides important insights into both the concerns about and the contributions of genetic technology. It assists in identifying factors that shape policy development related to genetic technology in general and genetic testing for late onset diseases in particular.

3. **Section three** indicates literature *specific to the topic of genetic testing for late onset diseases*. This literature deals with ethical, social, financial, psychological and other factors affecting policy development for genetic testing for late onset diseases within both the North American and European context.

All three sections contribute valuable information and important perspectives in analyzing and synthesizing the current literature related to genetic testing for late onset diseases.

General Bibliography

Durand, Guy. *Introduction générale à la bioéthique*. Fides & Cerf, 1999.

An important general introduction to bioethics from the francophone perspective. Addresses significant underlying issues that have implications for genetic technology in general and genetic testing for late onset diseases in particular.

Jonas, Hans. *Imperative of Responsibility: In Search of an Ethics for the Technological Age*. Chicago: University of Chicago Press, 1984.

Explores the importance of responsibility in a technological age — a responsibility to the future. Suggests an ethics of caution in the face of the potential damage of technology for future generations.

Lenoir, Noëlle, and Bertrand Mathieu. *Les norms internationales de la bioéthique*. Presses Universitaires de France, 1998.

Examines the relationship between ethics and the law in the international context. Elaborates on the international regulation of bioethics.

Lonergan, Bernard J. F. *Insight: A Study of Human Understanding*. 3rd ed. New York: Philosophical Library Inc., 1970.

An in-depth study of how people experience, understand, judge and make decisions. Identifies conditions or patterns that bring about either progress or decline. Identifies a scale of values from which one might assess issues such as genetic testing for late onset diseases.

Marange, Valérie. *La bioéthique: la science contre la civilisation?* Le Monde et Marabout, 1998.

Addresses the risks that the permanent and multiform evolution of knowledge poses. Asks whether ethics has the right to put a stop to the pursuit of knowledge.

Melchin, Kenneth R. "The Challenges of Technological Society for the Understanding of Christian Faith." *Défis présents et à venir de l'université / Future Challenges Facing Catholic Universities*. Jacques Croteau, ed. Ottawa: Saint Paul University, 1990, pp. 123–38.

Addresses the link between technology and human meaning.

Nelkin, D., and L. Tancredi. *Dangerous Diagnostics. The Social Power of Biological Information*. New York: Basic Books, 1989.

A study of the pervasiveness of diagnostic testing and the potential it offers institutions to classify, categorize and ultimately control individuals. Explores the ethical, social, and legal implications of cutting-edge technologies that can lead to new forms of discrimination in the name of standardized, objective measurements. Cautions against the creation of an underclass deemed unemployable, untrainable or uninsurable by such diagnostic tests.

Saint-Arnaud, Jocelyne. *Enjeux éthiques et technologies biomédicales*. Montréal: Presses de l'Université de Montréal, 1999.

The recent developments of biomedical technology have considerably modified, in the clinical setting, the implications of practice and the price of decision. In a context in which the ethical frontiers are in constant confusion, the establishment of solid landmarks is imposed. Poses that the framework of regulation continue between the new possibilities of technology and the principles that require a minimum supervision.

Schrecker, Ted, and Margaret A. Somerville. *Making Ethically Acceptable Policy Decisions: Challenges Facing the Federal Government*. London ON: Ted Schrecker Research and Consulting, 1997.

This report addresses the need for the National Biotechnology Strategy Interdepartmental Working Group to clarify what it means to say that an action, practice or policy is ethically acceptable. Also considers how the federal government should respond in situations when the answer is unclear, or when considerable conflict exists about the appropriate answer.

Tribe, Laurence H. "Technology Assessment and the Fourth Discontinuity: The Limits of Instrumental Rationality." *Southern California Law Review* 46 (1973), pp. 617–60.

Explores a manner of assessing technology in terms of technologies being treated as parts of ourselves rather than as mere tools or machines. Addresses the importance of enriching instrumental policy analysis concerning technologies with an understanding of technology as constitutive.

Genetic Technology

Altman, Scott. "(Com)modifying Experience." *Southern California Law Review* 65 (1991), pp. 293–340.

Focusses on the argument that technologies alter sensibilities and attitudes. Though altered sensibilities can lead to bad acts, the author posits that the changed attitudes and experiences are themselves harms. The article cautions against broad claims that technologies will alter everyone's experience. Making important decisions based on concern for preserving sensibilities is problematic.

Angier, Natalie. "Great 15-year Project to Decipher Genes Stirs Opposition" [available on the *New York Times* website: <http://search.nytimes.com/>]. *The New York Times*, June 5, 1990.

Discusses criticisms of and opposition to the Human Genome Project.

Annas, George J., and Sherman Elias, eds. *Gene Mapping: Using Law and Ethics as Guides?* New York: Oxford University Press, 1992.

Includes 17 chapters by U. S. authors focussing on the ethical, legal and social impact (ELSI) of the Human Genome Project. It defines many of the ELSI issues and makes a priority list in a U.S. context.

Appleyard, Bryan. *Brave New Worlds: Staying Human in the Genetic Future*. New York: Viking Press, 1998.

A user-friendly critique of genetic technology. The author addresses both the potential positive and negative consequences. Informative at a very basic level about ethical and social issues concerning genetic technology.

Baker, Robert. "A Theory of International Bioethics: Multiculturalism, Postmodernism, and the Bankruptcy of Fundamentalism," *Kennedy Institute of Ethics Journal* 8,3 (1998): 201–31.

———. "A Theory of International Bioethics: The Negotiable and the Non-Negotiable," *Kennedy Institute of Ethics Journal* 8, 3 (1998): pp. 233–73.

In both these articles, the author analyzes the justifiability of international bioethical codes. Are there certain "basic" or "fundamental" moral principles that are universally accepted in all cultures and eras? The first article assesses the challenges that multiculturalism and postmodernism pose to fundamentalism, and concludes that these challenges render the position philosophically untenable, thereby undermining the received conception of the foundations of international bioethics. The second

article offers an alternative model — a model of negotiated moral order — as a viable justification for international bioethics and for transcultural and transtemporal moral judgements.

Bertin, Joan E., and Mary S. Henifin. "Science, Law, and the Search for Truth in the Courtroom: Lessons from *Daubert v. Merrell Dow*." *The Journal of Law, Medicine and Ethics* 22,1 (1994): 6–20.

Deals with the relationship between science and law. Addresses the problem of trying to resolve questions of scientific uncertainty in the courtroom. Suggests that the law's traditional role of establishing normative behavioural standards can occur even in the absence of complete information.

Biesecker, Barbara Bowles. "Future Directions in Genetic Counseling: Practical and Ethical Considerations," *Kennedy Institute of Ethics Journal* 8, 2 (1998): 145–60.

Considers that certain ethical principles now guiding genetic counselling may have to be modified as genetic testing becomes more widespread in order to meet the changing needs of clients and society.

Boyle, P. J. "Shaping Priorities in Genetic Medicine." *Hastings Center Report* 25,3 (1995): 2–11.

Discusses whether society should make all or only some of the potential and existing genetic technologies widely available.

British Medical Association. *Human Genetics: Choice and Responsibility*. Oxford: Oxford University Press, 1998.

Assesses the ethical, legal and professional issues raised by human genetics in clinical practice. Confronts the potential for conflict between individuals' choices and their moral responsibilities to other people.

Brock, Dan W. "The Human Genome Project and Human Identity." *Houston Law Review* 29 (1992), pp. 7–22.

Addresses the effects that the Human Genome Project will most likely have on our conceptions of persons and of individuals' psychological sense of identity and the subsequent affect this will have on the law.

Caplan, A. L. "If Gene Therapy is the Cure, What is the Disease?" *Gene Mapping: Using Law and Ethics as Guides?* G. J. Annas and S. Elias, eds., 1992.

———. "The Concepts of Health and Disease." *Medical Ethics*. R. Veatch, ed., Boston: Jones and Bartlett, 1989, pp. 49–63.

Both articles are concerned with the subjectiveness of defining health and disease and the vulnerability of this enterprise to political and social influences.

Caulfield, Timothy A. *The Commercialization of Human Genetics: A Discussion of Issues Relevant to the Canadian Consumer*. A paper prepared for Industry Canada, Office of Consumer Affairs, 1997.

This paper addresses the many concerns regarding the commercialization of human genetics, largely from the perspective of the Canadian consumer, by reviewing the literature, commentaries, position papers and relevant law that touch these considerations.

———. "The Practice of Human Genetics: Emerging Areas of Consensus?" *Health Law Journal* 3 (1995): pp. 307–20.

Drawing on the conclusions of a three-year Canadian research project entitled Professional Norms in the Practice of Human Genetics, the article reviews both the emerging professional norms in the practice of human genetics and the potential issues surrounding these norms.

Centers for Disease Control. *Translating Advances in Human Genetics into Public Health Action: A Strategic Plan* [available online at <http://www.cdc.gov/genetics/publications/strategic.htm>]. Centers for Disease Control and Prevention, October 1, 1997.

Deals with how to use knowledge from genetics research to promote health and prevent disease and disability.

CIBA Foundation Symposium 149. *Human Genetic Information: Science, Law and Ethics*. Amsterdam: Elsevier North Holland, 1990.

A series of essays delivered at the CIBA foundation symposium.

Cook-Deegan, Robert. *The Gene Wars: Science, Politics, and the Human Genome*. New York: Norton, 1995.

A book about the politics of the Human Genome Project with insights into the project not only as science but also in terms of the development of government policies.

Council of Europe. "Convention for Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Biomedicine: Convention on Human Rights and Biomedicine." *Kennedy Institute of Ethics Journal* 7,3 (1997): 277–90.

Adopted November 19, 1996, and made available to countries to sign at a ceremony in Spain on April 4, 1997.

Cranor, Carl F. ed. *Are Genes Us?: The Social Consequences of the New Genetics*. New Brunswick NJ: Rutgers University Press, 1994.

Eleven essays dealing with ethical, legal, social and political consequences of genetics.

Cutter, Mary Ann G., et al. *Mapping and Sequencing the Human Genome: Science, Ethics, and Public Policy*. Colorado Springs: BSCS and the American Medical Association, 1992.

Considers the scientific, ethical and public policy issues surrounding the Human Genome Project.

Davis, Joel. *Mapping the Code: The Human Genome Project and the Choices of Modern Science*. New York: John Wiley & Sons, 1990.

Identifies the moral issues in the debate and reports on the impact of tensions on the scientific community.

Dommel, F. William Jr., and Duane Alexander. "The Convention on Human Rights and Biomedicine of the Council of Europe." *Kennedy Institute of Ethics Journal* 7,3 (1997): 259–76.

Describes the background and general provisions of the Convention on Human Rights and Biomedicine, which is the first international treaty focussed on bioethics. Also the Convention's requirements with those of U. S. federal regulations governing research with human subjects.

Dreyfuss, Rochelle Cooper, and Dorothy Nelkin. "The Jurisprudence of Genetics." *Vanderbilt Law Review* 45, 2 (1992): 313–48.

Focusses on the impact of genetic research on traditional legal concepts, in particular the impact of genetic research on core principles on which current norms are based. Law makers are compelled to reconsider the legal rules that mediate the relationships among persons and between individuals and the broader community.

Drlica, Karl A. *Double-Edged Sword: The Promises and Risks of the Genetic Revolution*. Helix/Addison Wesley, 1995.

Explores the positive and negative aspects of modern genetics. Discusses what the latest findings mean for those who suffer from genetic diseases as well as for those who want to use genetic screening to improve their lives.

Eisenberg, Rebecca. "Patents: Help or Hindrance to Technology Transfer." *Biotechnology*. Frederick B. Rudolph and Larry V. McIntyre, eds. Joseph Henry, 1996, pp. 161–74.

Deals with whether allowing patents on human genetic material will advance or impede scientific progress.

Espey, Jennifer, et al. *Socialethical Implications of Biotechnology*. Report sponsored by the Department of Western Economic Diversification. Envision Research, 1997.

Discusses the social implications of biotechnology to facilitate the development of the biotechnology industry. Clarifies the history of the policy debate, analyzes competing social and ethical perspectives of biotechnology, and discusses the competing pressures on government to address socioethical issues. Biotechnology is an opportunity for a public debate over how our society will develop more broadly. It is not the science of biotechnology that is in question; it is the manner in which specific applications of biotechnology will affect the way we live that is in debate.

Fisher, Lawrence M. “Profits and Ethics Clash in Research on Genetic Coding” [available on the *New York Times* website: <http://verify.nytimes.com/library/national/science/013094sci-genome.html>]. *The New York Times*. January 30, 1994.

Deals with the tension among scientists with respect to using information from the Human Genome Project for profit.

———. “The Race to Cash in on the Genetic Code” [available on the *New York Times* website: <http://verify.nytimes.com/library/financial/Sunday/082999invest-genomics.html>]. *The New York Times*. August 29, 1999.

Deals with the potential monetary payoff of the Human Genome Project and the dynamics that are involved as players and investors seek to be part of that payoff.

Fletcher, John C., and Dorothy C. Wertz. “Ethics, Law, and Medical Genetics: After the Human Genome is Mapped.” *Emory Law Journal* 39 (1990): pp. 747–809.

Given the inevitability of mapping the human genome, the authors question how this will affect the status of the specific duties of respect for and protection of the autonomous choices and privacy of persons, and respect for and protection of vulnerable human beings who are, or will become, incapacitated.

———. “Proposed: An International Code of Ethics in Medical Genetics Before the Human Genome is Mapped.” *Genetics, Ethics and Human Values: Human Genome Mapping, Genetic Screening and Therapy*, 1990.

Frankel, Mark S., and Albert Teich, eds. *The Genetic Frontier: Ethics, Law, and Policy*. Washington, D.C.: American Association for the Advancement of Science, 1994.

Deals with ethical, legal and policy issues related to genetic technology.

Genetics, Ethics and Human Values: Human Genome Mapping, Genetic Screening and Gene Therapy. Proceedings of the 24th CIOMS Round Table Conference, Tokyo and Inuyama City, Japan, July 22–27, 1990. Z. Bankowski and A. M. Capron, eds., Geneva: CIOMS, 1990.

Held under the auspices of the Science Council of Japan and co- sponsored by the World Health Organization and the United Nations Educational, Scientific and Cultural Organization.

Gert, Bernard, et al., eds. *Morality and the New Genetics: A Guide for Students and Health Care Providers*. Sudbury MA.: Jones and Bartlett Publishers, 1996.

An introduction to ethical dilemmas emerging from issues related to the work of the Human Genome Project. Deals with a variety of issues. Concerning genetic testing for late onset diseases, chapters 4 and 5 tackle the experience of Huntington disease, outlining the various challenges of this disease from a genetic point of view. The book also provides advice for health care professionals who work in situations involving genetic testing.

Haker, Hille, et al., eds. *Ethics of Human Genome Analysis: European Perspectives*. Tübingen: Attempto Verlag, 1993.

Several essays from the European context dealing with ethical issues surrounding the Human Genome Project.

Hanson, Mark J. "Religious Voices in Biotechnology: The Case of Gene Patenting," *Hastings Center Report* 27 (1997), special supplement, pp. 1–21.

The author shows that a key issue behind religious objections to patenting of life forms and genetic materials is how "commodification" will alter our understanding of them.

Heller, Jan Christian. *Human Genome Research and the Challenge of Contingent Future Persons. Toward an Impersonal Theocentric Approach to Value*. Omaha: Creighton University Press, 1996.

Investigates how the Human Genome Project is likely to affect future generations. Explores, from a theological perspective, the implications these effects hold for evaluating genetic research. Examines economic, political and philosophical issues underlying genetic research.

Holtzman, Neil A. *Proceed With Caution: Predicting Genetic Risk in the Recombinant DNA Era*. Baltimore: John Hopkins University Press, 1989.

Addresses the potential risks involved in the Human Genome Project.

Human Genetics Commission. *The UK Regulatory and Advisory Framework for Human Genetics*. London: Health Departments of the United Kingdom, 2000.

This document provides brief descriptions of the main bodies in the U.K. regulatory and advisory framework for human genetics, including current developments in their work when this is relevant to the work of the Human Genetics Commission.

Institute of Medicine, Committee on Assessing Genetic Risks. *Assessing Genetic Risks: Implications for Health and Social Policy*. L.B. Andrews, et al., eds., Washington: National Academy Press, 1994.

The Committee on Assessing Genetic Risks included basic researchers, medical geneticists, genetic counsellors, experts in law and ethics, and executives of the insurance industry. The committee was convened to discuss some of the moral and ethical issues involved in genetic testing. Several policy recommendations emerge from the publication.

Kaveny, M. Cathleen. "Jurisprudence and Genetics." *Theological Studies* 60 (1999), pp. 135–47.

The author grapples with the proper relation of law and morality to three emerging issues connected with genetics: cloning, discrimination on the basis of genetic information, and patenting of genetic material.

Keenan, James. "Genetic Research and the Elusive Body." *Embodiment, Morality, and Medicine*. Lisa S. Cahill and Margaret A. Farley, eds. Dordrecht, Netherlands: Kluwer Academy, 1995, pp. 59–73.

Argues against a Platonic/Cartesian dualism that sees the body as a res extensa with no relation to our human nature or our person. Reasons that a separation misunderstands personhood. Demonstrates the necessity of keeping the body-person at the centre of ethical analysis.

Kevles, Daniel J. "Vital Essences and Human Wholeness: The Social Readings of Biological Information." *Southern California Law Review* 65 (1991), pp. 255–78.

Addresses the dialectical tension between the reductionist and expansionist arguments concerning the human person. The Human Genome Project will provide enormous information concerning biological factors shaping human life; however, the author argues, this cannot eclipse the transcendent wholeness of a human person.

Kevles, Daniel J. and Leroy Hood (eds.). *The Code of Codes: Scientific and Social Issues in the Human Genome Project*. Cambridge MA: Harvard University Press, 1992.

Fourteen essays exploring the substance and possible consequence of the Human Genome Project in relation to ethics, law and society.

Kilner, John, et al., eds. *Genetic Ethics: Do the Ends Justify the Genes?* Grand Rapids: Wm. B. Eerdmans, 1997.

Addresses, from a Christian perspective, the ethical questions and difficult personal and social decision-making situations raised by rapid genetic advancements. The chapters deal with various dimensions of the genetic challenge: genetic perspective, genetic information and genetic intervention.

Kimyai-Asadi, A., and P. B. Terry. "Ethical Considerations in Pulmonary Genetic Testing and Gene Therapy." *American Journal of Respiratory and Critical Care Medicine* 155,1 (1997): 3–8.

Deals with medical ethics, gene therapy, genetic screening and lung diseases.

Lenoir, Noëlle. "UNESCO, Genetics, and Human Rights." *Kennedy Institute of Ethics Journal* 7,1 (1997): 31–42.

In response to a mandate conferred on the International Bioethics Committee (IBC) of UNESCO in November 1993, the IBC drafted a "universal declaration on the human genome and human rights," which was considered by the General Conference of UNESCO in November 1997. This article discusses the development of the document and provides the text of the "revised preliminary draft" of the declaration.

Lippman, A. "Prenatal Genetic Testing and Screening: Constructing Needs and Reinforcing Inequities." *American Journal of Law and Medicine* 17 (1991), pp. 15–50.

Concerned with the subjective element in defining health and disease and the vulnerability of this enterprise to political and social influences.

Macer, Darryl R. J. *Bioethics for the People by the People* [available online at <http://www.biol.tsukuba.ac.jp/~macer/BFP.html>]. Tsukuba, Japan: Eubios Ethics Institute, 1994.

Results of a 1993 survey performed across 10 countries of the world (Australia, Hong Kong, India, Israel, Japan, New Zealand, the Philippines, Russia, Singapore and Thailand) are compared to the results of surveys conducted in North America and Europe. The focus of the survey was to understand how people from diverse cultures and geographical locations think about bioethics. Specifically, looks at how people think about diseases, life, nature and selected issues of science and technology, biotechnology, genetic engineering, genetic screening and gene therapy. The book also includes a series of papers on international bioethics from different countries of the world, representing academic approaches and descriptions of these issues. Three titles of importance in the context of genetic testing for last onset diseases are Michael S. Yesley, "Trends in Bioethics in the United States," pp. 41–45, Paul R. Billings, "Genetic Information in the Health Care Reform Era," pp. 51–56, and Judge Christian Byk, "Bioethics Within the Council of Europe: The Protection of Genetic Information," pp. 68–73.

———. "Whose Genome Project?" *Bioethics* 5 (1991), pp. 183–211.

Addresses the issue of ownership of the Human Genome Project.

Marteau, Theresa, and Martin Richards, eds. *The Troubled Helix: Social and Psychological Implications of the New Human Genetics*. New York: Cambridge University, 1996.

A series of essays from a British perspective in regard to genetic testing.

Mauron, Alex. "La génétique et le souci éthique des générations futures." *Folia Bioethica* 14 (1993).

Wrestles with the intergenerational relationship and the responsibility of the present generation to the future generation especially in relation to genetic research.

McTeer, Maureen A. "A Role for Law in Matters of Morality," *McGill Law Journal* 40 (1995), pp. 894–903.

Examines whether there exists a role for law in moral matters. Considers this in relation to genetic technologies. Favours a public interest approach to reproductive and genetic technologies because of the far-reaching impact of the technologies and because of the law's role in maintaining public order.

Murphy, Timothy F., and Marc A. Lapp, eds. *Justice and the Human Genome Project*. Berkeley: University of California Press, 1994.

Nine essays probing the potential social uses and abuses of detailed genetic information. Addresses ethical, legal and social implications of genetic research.

Murray, Thomas H. "Assessing Genetic Technologies: Two Ethical Issues." *International Journal of Technology Assessment in Health Care* 10,4 (1994): 573–82.

Although focussed on reproductive technology, this article is useful in terms of genetic testing for late onset disease in its discussion of 10 factors that characterize the social context of contemporary genetics and that are important in understanding the implications of genetic technology.

Murray, Thomas H., et al., eds. *The Human Genome Project and the Future of Health Care*. Bloomington and Indianapolis: Indiana University, 1996.

This book examines how the Human Genome Project will alter the shape and content of American health care. Leading scholars explore the clinical, ethical, legal and policy implications of the project to see how it may affect the forms of health care available, who delivers it, who receives it and who pays for it.

National Institutes of Health-Department of Energy Working Group on Ethical, Legal, and Social Implications of Human Genome Research. *Genetic Information and Health Insurance: Report of the Task Force on Genetic Information and Insurance*, Publication No. 93-3686. Bethesda, MD: National Institutes of Health, 1993.

Deals with the relationship between genetic information and health insurance.

Peters, Ted. *Genetics: Issues of Social Justice*. New York: Pilgrim Press, 1998.

Eleven chapters discussing genetic research in terms of social questions such as mass non-voluntary genetic screening of newborns, denial of insurance or employment based on knowledge of future disease, discarding of defective pre-embryos, and parents' selection of genes for positive traits. Looks at the social and religious implications of genetic research.

Resnik, David B. "The Morality of Human Gene Patents." *Kennedy Institute of Ethics Journal* 7,1 (1997): 43–61.

Discusses the morality of patenting human genes and genetic technologies. Examines arguments on the various sides of the issue. Highlights the need for a continual re-examination of genetic laws and policies.

Rothstein, Mark A., ed. *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era*. New Haven: Yale University, 1997.

Because the techniques for identifying human genes are so much more advanced than the abilities to alter them, the immediate challenges that the Human Genome Project presents for policy makers pertain to the control of genetic information. This volume addresses this question. It consists of a series of essays providing a comprehensive exploration of ethical, legal and social issues emerging in respect to advances in genetic research and issues of privacy and confidentiality.

Roy, David J., et al. *Bioethics in Canada*. Scarborough: Prentice Hall Canada Inc., 1994.

Situates the Human Genome Project historically, sketches the scientific developments leading up to the project, and reviews the evolution of ethical concern that has followed closely upon the heels of these developments. Raises social and ethics issues in relation to presymptomatic diagnosis and screening.

Sarkar, Sahotra. *Genetics and Reductionism*. Cambridge Studies in Philosophy and Biology. Cambridge: Cambridge University Press, 1998.

Questions and clarifies the meaning of genetic. Shows how molecular studies have affected genetics and provides the philosophical background necessary to understand the debates over the Human Genome Project.

Schrecker, Ted, et al. *Biotechnology, Ethics and Government*. Report to the Interdepartmental Working Group on Ethics. Montréal: Westminster Institute for Ethics and Human Values, McGill Centre for Medicine, Ethics and Law, 1997.

Considers the importance of an ethical perspective on public policy. Exclusion of ethics from policy analysis defers judgments to the workings of power and influence within the political process. The principle aim of the report is to vindicate the need for government to respond to the manifold ethical issues raised by biotechnology, and to direct that response in ways that are likely to produce responsible procedures and responsible outcomes.

Second National Conference on Genetics, Religion and Ethics. *Implications of the Human Genome Project for Medicine, Theology, Ethics and Policy*. March 13–15, 1992. Houston, Texas.

A series of essays that consider the implications of the Human Genome Project for medicine, theology, ethics and policy.

Shannon, Thomas A. "Ethical Issues in Genetics." *Theological Studies* 60 (1999), pp. 111–23.

A review of ethical issues in genetics through the lenses of privacy-confidentiality, risk-benefit analysis in relation to prenatal diagnosis and gene therapy, and freedom-determinism/human dignity in the context of cloning. The author provides an overview of developments in genetics and highlights thematic issues common to these developments.

Sherry, Stephen F. "The Incentive of Patents." *Genetic Ethics*. John F. Kilner, Rebecca D. Pentz, and Frank E. Young, eds. Grand Rapids: Eerdmans, 1997, pp. 113–23.

The author addresses the fact that the U.S. Patent and Trademark Office has indicated that the Thirteenth Amendment to the Constitution would prohibit patenting transgenic human persons (but this prohibition would likely not apply to embryos).

UNESCO. *Universal Declaration on the Human Genome and Human Rights* [available at <http://www.umn.edu/humanrts/instree/Udhrhg.htm>].

Walter, James J. "Theological Issues in Genetics." *Theological Studies* 60 (1999), pp. 124–34.

An analysis and evaluation of moral debates about issues in modern genetics and reproductive medicine from a theological perspective. The author appeals to two hermeneutical themes — human beings as "images of God" and the tendency of humans to "play God" — in order to discuss various church statements and theological literature on human gene transfer, somatic cell nuclear transplant cloning of human beings, and patenting of human genes.

Weir, Robert, et al., eds. *Genes and Human Self-Knowledge: Historical and Philosophical Reflections on Modern Genetics*. Iowa City: University of Iowa Press, 1994.

Essays discussing genetic technology and human identity from historical and philosophical perspectives.

Wertz, D. C. "Is There a "Women's Ethic" in Genetics: A 37-nation Survey of Providers." *Journal of the American Medical Women's Association* 52,1 (1997): 33–38.

Through a survey of 2,903 geneticists in 37 nations, compares the differences between women and men in dealing with ethical questions concerning genetics.

Wilkie, Tom. *Perilous Knowledge: The Human Genome Project and Its Implications*. Berkeley: University of California Press, 1993.

British physicist and writer outlines the history of the Human Genome Project. He considers the potential positive and negative social consequences.

World Health Organization. *Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services*. Report of a WHO meeting on ethical issues in medical genetics, Geneva, December 15–16, 1997 [available on the Internet at <http://www.who.org/ncd/hgn/hgnethic.htm>].

This document is the result of an original draft that was circulated worldwide; comments were received from all regions and WHO staff. This formed the background of the WHO meeting. The participants were experts in this field from both developing and developed countries.

World Medical Association Declaration of Helsinki. *Recommendations Guiding Physicians in Biomedical Research Involving Human Subjects* [available on the Internet at <http://business.virgin.net/cdss.com/Declaration.htm>]. 1996.

Genetic Testing for Late Onset Diseases

Ad Hoc Committee on Genetic Testing/Insurance Issues. "Genetic Testing and Insurance." *American Journal on Human Genetics* 56,1 (1995): 327–31.

Addresses complex questions about the appropriate relationship among providers of genetic testing, patients and insurers in the U.S. The authors maintain that until universal access to health care is a reality, genetic testing and genetic diagnosis will raise important issues for the practising geneticist. How much does a client need to know about insurance implications before consenting to a genetic test? Should patients be counselled to purchase insurance before being tested? Should genetic information be excluded from medical records before their release to insurance companies for routine reimbursements or underwriting? What are the ethical and legal responsibilities of the geneticist?

Advisory Committee on Genetic Testing. *Advice to Research Ethics Committees: Points to Consider in Ethical Review of Medical Research Involving Genetic Testing* [available on the Internet at <http://www.doh.gov.uk/genetics/recrev3.htm>]. London: Health Departments of the United Kingdom, 1998.

Sets out in the form of questions to researchers issues that research ethics committees may wish to consider before giving ethical approval to research that includes genetic testing.

———. *Genetic Testing for Late Onset Disorders*. London: Health Departments of the United Kingdom, 1998.

Sets out the issues to be considered before genetic testing for late onset disorders is offered and during the provision of such tests. Major issues relate principally to requests for genetic testing from healthy relatives of patients with a late onset genetic disorder.

Allan, David. "Ethical Boundaries in Genetic Testing." *Canadian Medical Association Journal* 154,2 (1996): 241–44.

Looks at ethical principles that should guide decisions about genetic testing, and the importance of communicating these principles to patients and their families.

Alper, Joseph S. "Does the ADA Provide Protection Against Discrimination on the Basis of Genotype?" *Journal of Law, Medicine, and Ethics* 23 (1995), pp. 167–72.

Presents detailed arguments justifying the position that the Americans with Disabilities Act provides a legal remedy for a presently asymptomatic individual who, as the result of genetic tests, is found to have the genotype for a multifactorial disease and, consequently, suffers discrimination.

American Medical Association, Council on Ethical and Judicial Affairs. "Multiplex Genetic Testing." *Hastings Center Report* 28,4 (1998), pp. 15–21.

Addresses the implications that result from various kinds of genetic testing. In particular, genetic tests raise concerns about informed consent and they pose various needs for counselling.

American Society of Human Genetics. "American Society of Human Genetics Statement on Genetic Testing for Breast and Ovarian-Cancer Predisposition." *American Journal of Human Genetics*, 55,5 (1994): i–iv.

American Society of Human Genetics Board of Directors/American Council of Medical Genetics Board of Directors. "Points to Consider: Ethical, Legal and Psychological Implications of Genetic Testing in Children and Adolescents." *American Journal of Human Genetics*, 57 (1995), pp. 1233–41.

Considers the ethical, legal and psychological implications of genetic testing in children and adolescents.

Andrews, Lori B., and Ami S. Jaeger. "Confidentiality of Genetic Information in the Workplace." *American Journal of Law and Medicine* 17,1&2 (1991): 75–108.

Analyzes existing legal protections for the confidentiality of information collected through genetic screening or genetic monitoring in the workplace. Analyzes legal bases for employee and third-party access to the employee's genetic information. Given gaps in existing legal protections, suggests parameters for a model law protecting the confidentiality of genetic information collected in the workplace.

Annas, George J., Leonard H. Glantz, and Patricia A. Roche. "The Genetic Privacy Act and Commentary" [available on the Internet at <http://www.bumc.bu.edu/www/sphlw/gpa>].

The authors of the act describe its purpose in "The Genetic Privacy Act: A Proposal for National Legislation." Jurimetrics 37 (1996), p. 1–11.

Annas, George J., Leonard H. Glantz, and Patricia A. Roche. "Drafting the Genetic Privacy Act: Science, Policy, and Practical Considerations." *The Journal of Law, Medicine and Ethics* 23 (1995), pp. 360–66.

The drafting of the Genetic Privacy Act began as an effort to develop rules for gene banks. It developed into an awareness that more is needed. It was suggested that federal legislation is needed to protect individual privacy by protecting not only stored DNA samples but also the genetic information obtained from analyzing DNA samples. The core of the Act prohibits individuals from analyzing DNA samples unless they have verified that written authorization for the analysis has been given by the individual or the individual's representative.

Benowitz, Steven. "Scientists Struggling With Concerns Raised by Genome Project Progress." *The Scientist* 10,14 (1996):1, 6–7.

Report indicating that many scientists are finding that concerns about the complex ethical, legal and social issues surrounding genetic testing and the use of the resulting information are taking up a larger part of their time. Also that some firms and institutions are establishing ethics branches to focus on policy issues left unresolved by National Institutes of Health's Ethical Legal and Social Issues project.

Bove, C. M., et al. "Presymptomatic and Predisposition Genetic Testing: Ethical and Social Considerations." *Seminars in Oncology Nursing* 13,2 (1997): 135–40.

An overview of the ethical and social concerns that are raised by the use of new genetic tests in asymptomatic persons. It draws on review articles, research studies and legislation related to genetic testing. The study draws some insightful conclusions and suggests the importance of more research being done to identify the family concerns, and to develop effective educational, counselling and supportive interventions.

Braude, P. R., et al. "Non-Disclosure Preimplantation Genetic Diagnosis for Huntington's Disease: Practical and Ethical Dilemmas." *Prenatal Diagnosis* 18,13 (1998): 1422–26.

Deals with some of the practical and ethical problems that arise due to prenatal testing for Huntington disease.

Bridge, Peter. *Genetic Testing for Predispositions and Adult-Onset Disorders in Canada*. Report written for Health Canada, 1999.

Report on results of a small survey of laboratories in Canada conducting genetic tests for predispositions and adult-onset disorders. Also contains some background information and recommendations.

Brunger, Fern, and Ken Bassett. *Culture and Genetic Testing: Building a Research Program*. Report prepared for the Medical, Ethical, Legal and Social Issues Advisory Committee of the Canadian Genome Analysis and Technology Programme, 1997.

Explores the relationship between culture and genetic testing. Considers how both mutually define and transform the other, and highlights the need for a heightened awareness of this interweaving between culture and genetic testing.

Burgess, M. M. "Ethical Issues in Genetic Testing for Alzheimer's Disease: Lessons from Huntington's Disease." *Alzheimer Disease and Associated Disorders* 8,2 (1994): pp. 71–78.

Discusses an ethics research project in predictive testing for Huntington disease and the relevance of the findings for genetic testing for Alzheimer disease.

Calender, A., et al. “Recommandations ethiques dans le cadre du depistage genetique des affections endocrines a caractere hereditaire.” *Annales d’Endocrinologie* 58,4 (1997): 343–48.

Discusses recommendations about French laws published in July 1994, and analyzes their concrete applications in clinical and genetic follow-up of patients and family members defined as gene carriers in predisposed families.

Caulfield, T. A. “The Law, Adolescents, and the APOE Epsilon 4 Genotype: A View from Canada.” *Genetic Testing* 3,1 (1999): 107–13.

Discusses the testing of minors for APOE status to minimize the costs of participation in high-contact sports, such as ice hockey, soccer, rugby and boxing, in which head injury is likely to occur. Under some circumstances, testing minors for this purpose might be justifiable. The author states that this would raise complex ethical and legal questions about the respective powers and rights of adolescents and parents.

Chambers, Donald C. “Genetic-testing Issues Pose Biggest Test for Insurers.” *Reinsurance Reporter* [available on the Internet at <http://www.Inre.com/ep/rr/issues/143/143_4.htm>], 1999.

A speech delivered during the session on bioethics and insurance at the Reavie Fifth International Life and A&H Insurance and Reinsurance Meeting in Cannes, France, October 1999. Proposes that genetic testing poses the most threatening underwriting issues of the century, with the potential to destroy the private insurance industry worldwide.

Cohen, Cynthia B. “Wrestling with the Future: Should We Test Children for Adult-Onset Genetic Conditions?” *Kennedy Institute of Ethics Journal* 8,2 (1998): 111–30.

Suggests that weighing the harms and benefits of testing children for late onset diseases results in a draw, with no substantial harms proven. Suggests that, in fact, testing may enhance rather than violate the adult autonomy of tested children.

Collins, Francis S. “BRCA1 — Lots of Mutations, Lots of Dilemmas.” *New England Journal of Medicine* 334 (1996), pp. 186–88.

Suggests positive strategies for utilizing genetic information on breast cancer.

Collins, Francis S. “Shattuck Lecture — Medical and Societal Consequences of the Human Genome Project.” *New England Journal of Medicine* 341,1 (1999), pp. 28–37.

Discusses the Human Genome Project in relation to diagnosis of hereditary diseases and various social and ethical issues.

Cordi, A. M., and J. Brandt. “Psychological Cost and Benefits of Predictive Testing for Huntington’s Disease.” *American Journal of Medical Genetics* 55 (1995), pp. 618–25.

Discusses the psychological impact of genetic testing for late onset diseases.

Dickens, B. M. “Legal Issues in Predictive Genetic Testing Programs.” *Alzheimer Disease and Associated Disorders* 8,2 (1994): 94–101.

Reviews aspects of predictive genetic testing to which the general law of doctor-patient relations applies and identifies peculiarities of such testing that raise more specialized legal issues.

Dickens, B. M., et al. “Legal and Ethical Issues in Genetic Testing and Counseling for Susceptibility to Breast, Ovarian and Colon Cancer.” *Canadian Medical Association Journal* 154,6 (1994): 813–18.

From a Canadian perspective, addresses various legal and ethical issues in genetic testing for susceptibility to breast, ovarian and colon cancer. In particular, looks at the legal distinction between a breach of confidentiality and the legitimate sharing of information in a patient’s interest or to prevent harm to a third party. Speaks about the particular importance that feminist ethics may play in addressing these issues.

Dudokde Wit, A. C., et al. “Distress in Individuals Facing Predictive DNA Testing for Autosomal Dominant Late-Onset Disorders: Comparing Questionnaire Results with Indepth Interviews.” Rotterdam Leiden Genetics Workshop. *American Journal of Medical Genetics* 75,1 (1998): 62–74.

Discusses reactions and coping strategies of risk carriers for various late onset diseases.

———. “Predicting Adaptation to Presymptomatic DNA Testing for Late-Onset Disorders: Who Will Experience Distress?” Rotterdam Leiden Genetics Workshop. *Journal of Medical Genetics* 35,9 (1998): 745–54.

Report on the first comparative study on predicting post-test distress following presymptomatic genetic testing for Huntington disease, cancer syndromes and hereditary breast and ovarian cancer.

Eaton, M. L.. “Surrogate Decision Making for Genetic Testing for Alzheimer Disease.” *Genetic Testing* 3,1 (1999): 93–97.

Deals with the ethical and legal problems posed by the commercial availability of genetic tests for the purpose of aiding in the differential diagnosis of Alzheimer disease among patients with dementia. Such patients often lack the mental capacity to consent to or reject such testing. If genetic testing is to be undertaken, it is important legally and ethically to consider who should participate in the decision to test. Generally, according to law in the United States, the patient’s previously expressed wishes determine which individual should serve as the surrogate decision maker. The author feels that other family members should be included in the discussion of the decision, and their assent to the surrogate’s decision should be sought.

Elger, B. S., and T. W. Harding. “Testing Adolescents for a Hereditary Breast Cancer Gene (BRCA1): Respecting their Autonomy is in Their Best Interest.” *Archives of Pediatrics and Adolescent Medicine* 154,2 (2000): 113–19.

Argues that there are not enough best interest arguments to deny genetic testing to an adolescent at risk of carrying a BRCA1 mutation, even when the competence of adolescents for medical decisions is considered to be lower than the competence of adults.

Feingold, J., and A. Durr. “Le diagnostic presymptomatique en genetique: le modèle de la maladie de Huntington.” *Pathologie Biologie* 45,3 (1997): 209–12.

Indicates the ethical problems predictive testing of late onset hereditary disorders raises, especially related to the severity of the disease and its absence of treatment.

Flanagan, W. F. “Genetic Data and Medical Confidentiality.” *Health Law Journal* 3 (1995), pp. 269–88.

Considers the extent to which current laws and practices can be appropriately applied to genetic data, and questions whether common law precedent can provide a useful and accurate indication of how the law may develop as the use of genetic data grows. The objective of the article is to provide a helpful and practical guide for patients, professionals and policy makers in the area of the confidentiality of genetic data.

Fraser, F. C. “Some Complexities in the Application of Guidelines for Genetic Testing.” *Health Law Journal* 3 (1995), pp. 301-6.

Argues that the genetic counselling process varies greatly at the various stages of genetic testing, and that ethical guidelines for its practice should be interpreted with this in mind.

Freedman, T. G. “Genetic Susceptibility Testing: Ethical and Social Quandaries.” *Health and Social Work* 23,3 (1998): 214–22.

In the U.S. context, as health care places more emphasis on illness prevention and simultaneously commits less economic support, genetic testing presents social and ethical challenges as well as dilemmas. Based on a study consisting of intensive field observation and in-depth face-to-face interviews concerning genetic susceptibility testing. The author asserts that the social worker may be in a unique position to collaborate with other health professionals in the clinical and the policy arena in regard to these tests.

Fucci, S.“Biotechnologies and Predictive Medicine: Legal Aspects.” *Forum: Trends in Experimental and Clinical Medicine*. Proceedings 9.3, suppl. 3 (1999), p. 88–92.

The author examines problems incurred by the development of predictive medicine in light of ethical citations and guidelines, included in the following policy initiatives: the National Committee for Bioethics in Gene Therapy, 1991; the Italian privacy law, 1996; the European Convention on Bioethics, 1996; the 98/44/CE Directive on the Patentability of Biotechnological Inventions; and the Deontological Mecial Code, 1998.

Fujimura, F. K. “Diagnosis and the New Genetics.” *Current Opinion in Biotechnology* 5,6 (1994): 654–62.

While acknowledging the many potential benefits of the transfer of molecular technology to the area of clinical genetic analysis, the author raises concerns regarding the possible misuse of genetic tests and information, particularly with regard to presymptomatic diagnosis of disease and population screening.

Garber, J. E., and A. F. Patenaude. “Ethical, Social and Counselling Issues in Hereditary Cancer Susceptibility.” *Cancer Survey* 25 (1995), pp. 381–97.

Deals with the newness of many issues to the medical community and to the public that genetic testing for late onset disease raises. Concerned that the “explosive technology” be used responsibly so that the potential benefits of genetic knowledge are not eclipsed by the risks to autonomy, privacy and justice. Addresses the considerable effort already under way in the U.K., Europe and the U.S. at the research and social levels to create protection for individuals found to carry genetic susceptibility to disease.

Geller, G., and N. A. Holtzman. “A Qualitative Assessment of Primary Care Physicians’ Perceptions About the Ethical and Social Implications of Offering Genetic Testing.” *Quality Health Research* 5,1 (1995): 97–116.

Addresses the impact of genetic testing from the perspective of primary care physicians.

Gevers, J. K. M. “Genetic Testing and Insurance.” *Medicine and Law* 11 (1992), pp. 541–545.

Addresses the public debate in the Netherlands concerning the extent to which insurance companies should be allowed to collect genetic data on persons seeking insurance coverage. Discusses arguments underlying the temporary agreement reached between insurers and the government and in particular the question of whether genetic information should have the same status under the law as other medical information.

Godard, B., et al. “Ethical Issues in Establishing a Registry for Familial Alzheimer’s Disease.” *Alzheimer Disease and Associated Disorders* 8,2 (1994): 79–93.

Examines initial considerations of the field work involved in developing a registry pertaining to genetic testing for Alzheimer disease and familial Alzheimer disease by linkage analysis.

Gostin, Lawrence O. “Genetic Privacy.” *The Journal of Law, Medicine and Ethics* 23 (1995), pp. 320–30.

Examines a particular aspect of health information — genetic privacy. Identifies a conceptual structure relating to the acquisition and use of genomic information.

Grady, C. "Ethics and Genetic Testing," *Advanced Internal Medicine* 44 (1999), pp. 389–411.

Addresses the serious issues for individuals and society related to the circumstances under which genetic information should be sought and the uses that should be make of such information. Suggests that ethical reflection and analysis will help society to prepare for the responsible use of information about genotypes so that individuals benefit, justice is served, and confidentiality, privacy, respect for autonomy, dignity and differences of each individual are preserved.

Hall, Mark A., and Wendy R. Uhlmann. "When Genes are Decoded, Who Should See the Results?" [available on the *New York Times* website: <http://search.nytimes.com/>]. *The New York Times*, February 29, 2000.

Comment on the need for safeguards in light of the growth of the possibilities for using genetic tests to help identify people with increased risk of developing certain diseases.

Harris, John. "The Principles of Medical Ethics and Medical Research." *Cadernos de Saúde Pública / Reports in Public Health* 15,1 (1999): 7–13.

Discusses the application of the principles of medical ethics and of medical research. The author considers the particular problems created by research and predictive testing on children for late-onset conditions and goes on to discuss research on those whose consent is problematic more generally. The author makes recommendations for the reform of The Declaration of Helsinki and of the International Ethics Guidelines for Biomedical Research Involving Human Subjects.

Holtzman, Neil A., and Michael S. Watson, eds. *Promoting Safe and Effective Genetic Testing in the United States*. Final report of the Task Force on Genetic Testing. The National Human Genome Research Institute, 1997.

The final report of an intensive study undertaken by the Task Force on Genetic Testing, which was created by the National Institutes of Health-Department of Energy Working Group on Ethical, Legal, and Social Implications of Human Genome Research. The mandate of the Task Force was to review genetic testing in the United States and make recommendations to ensure the development of safe and effective genetic tests. The prime concern of the Task Force was predictive uses of genetic tests performed in healthy or apparently healthy people. The report does not recommend policies for specific tests but suggests a framework for ensuring that new tests meet criteria for safety and effectiveness before they are unconditionally released.

Holtzman, S., et al. "Decision Analysis and Alzheimer Disease: Three Case Studies." *Genetic Testing* 3,1 (1999): 71–83.

Deals with the usefulness of the analytic tool known as "decision analysis" in three specific cases of people dealing with Alzheimer disease. Of specific interest is the use of this tool for genetic testing for late onset of the disease.

Hubbard, Ruth, and Richard C. Lewontin. "Pitfalls of Genetic Testing." *New England Journal of Medicine* 334,18 (1996): 1192–93.

A highly critical view of genetic testing.

Juengst, E. T. "Caught in the Middle Again: Professional Ethical Considerations in Genetic Testing for Health Risks." *Genetic Testing* 1,3 (1997–98): 189–200.

Deals with questions about the predictive significance of particular genotypes. Considers ethical considerations that are becoming important to professional decision making about genetic testing: the psycho-social impact of testing, the patient's privileges with respect to testing, and the potential for effective prevention following testing.

Kadlec, J. V., and R. A. McPherson. "Ethical Issues in Screening and Testing for Genetic Diseases." *Clinics in Laboratory Medicine* 15,4 (1995): 989–99.

Addresses the ethical issues that will confront patients, physicians and laboratory personnel as genetic testing advances. Suggests that the primary ethical issues will focus on who has the right to request or compel genetic testing, who has access to confidential information, and what medical or social actions may be predicted legally on genetic information.

Kash, K. M. "Psychosocial and Ethical Implications of Defining Genetic Risk for Cancers." *Annals of the New York Academy of Science* 768 (1995), p. 41–52.

The author argues for the importance of providing fully informed consent regarding the hazards and the benefits of genetic testing and defining risk. The article outlines a model based on the ethical principles of autonomy, beneficence and confidentiality.

Kinder, B. K. "Genetic and Biochemical Screening for Endocrine Disease: II. Ethical Issues." *World Journal of Surgery* 22,12 (1998): 1208–11.

Looks at the need for a fundamental understanding of the ethical issues involved in the development of appropriate societal regulations regarding genetic screening.

Lancaster, J. M., et al. "An Inevitable Dilemma: Prenatal Testing for Mutations in the BRCA 1 Breast-Ovarian Cancer Susceptibility Gene." *Obstetrics and Gynecology* 87,2 (1996): 306–9.

Addresses the difficulty of formulating straightforward guidelines regarding prenatal BRCA1 testing and the importance of clinicians and health care providers being familiar with the nuances of the debate.

Lemmens, Trudo, and Poupak Bahamin. *Genetics in Life, Disability and Additional Health Insurance in Canada: A Comparative Legal and Ethical Analysis*. Report submitted to the Medical, Ethical, Legal and Social Issues Advisory Committee of the Canadian Genome Analysis and Technology Programme.

Seeks to clarify the role that genetic information might play in health insurance in Canada. Considers if and how genetic information differs from other health information already used by insurers. Considers how underlying views on justice affect the debate on the use of genetics for insurance purposes. Suggests that the human rights structures already in place in Canada could be used to promote (or function as a model for) a fair use of genetic information.

Lennox, A., et al. “Molecular Genetic Predictive Testing for Alzheimer’s Disease: Deliberations and Preliminary Recommendations.” *Alzheimer Disease and Associated Disorders* 8,2 (1994): 126–47.

Report of a workshop on genetic predictive testing for familial Alzheimer disease held in Toronto in 1993. Legal, ethical, biomedical and psychosocial issues related to establishing predictive testing programs for Alzheimer disease were discussed. A comparison is made between recommendations contained in the Ethical Issues Policy Statement for Huntington disease and the applicability of this to Alzheimer disease.

Leonard, D. G. “The Future of Molecular Genetic Testing.” *Clinical Chemistry* 45,5 (1999): 726–31.

Addresses the influence our understanding of the human genome, technological advances, and social, ethical and legal factors surrounding genetic testing has on how genetic information is translated into medical application. The author maintains that with time, new genetic information will be translated in clinical tests for the diagnosis of current illness and prediction of future disease risk, and will be used for the development of genetically directed therapies and preventive intervention. He predicts that most genetic testing will eventually be highly automated, with only rare genetic disease tests performed manually. Thus, the challenge for the clinical genetic laboratory will be to keep pace with this information explosion to provide state-of-the-art genetic testing and to ensure that the genetic test results are used in a morally, ethically and socially responsible way.

Lessick, M., and S. Faux. “Implications of Genetic Testing of Children and Adolescents.” *Holistic Nursing Practice* 12,3 (1998): 38–46.

Examines developmental theories concerning children’s ability to make choices, as well as informed consent and ethical considerations in genetic testing.

Mahowald, M. B. “Genetic Counseling: Clinical and Ethical Challenges.” *Annual Review of Genetics* 32 (1998), pp. 547–59.

Looks at the impact of the Human Genome Project on genetic counselling in light of the availability of presymptomatic tests for late onset disorders and the possibility of preventive behaviour or treatment.

Marsick, R., et al. “Genetic Testing for Renal Diseases: Medical and Ethical Considerations.” *American Journal of Kidney Disease* 32,6 (1998): 934–45.

Reviews renal diseases with a known genetic basis and the current methods available for genetic testing. Examines the potential medical indications for genetic testing and reviews the ethical considerations regarding genetic testing for renal diseases, recent genetic privacy legislation, and the special role genetic testing may have in transplantation.

McConnell, L. M., et al. “Evaluation of Genetic Tests: APOE Genotyping for the Diagnosis of Alzheimer Disease.” *Genetic Testing* 3,1 (1999): 47–53.

Assesses the recent clinical studies concerning APOE genotyping and develops a formal framework for evaluating its usefulness. The authors suggest that APOE genotyping presents foreseeable, significant psychosocial consequences for family members that must be weighed against any psychosocial benefit.

———. “Genetic Testing and Alzheimer Disease: Recommendations of the Stanford Program in Genomics, Ethics, and Society.” *Genetic Testing* 3,1 (1999): 3–12.

The multidisciplinary Alzheimer Disease Working Group of the Stanford Program in Genomics, Ethics, and Society presents comprehensive recommendations on genetic testing for Alzheimer disease. The group concludes that under current conditions genetic testing for Alzheimer disease prediction or diagnosis is only rarely appropriate. Although criteria for judging the readiness of a test for introduction into routine clinical practice typically relies heavily on evaluation of technical efficacy, the group recommends a broader and more comprehensive approach. This article outlines these recommendations.

McKinnon, W. C., et al. “Predisposition Genetic Testing for Late-Onset Disorders in Adults. A Position Paper of the National Society of Genetic Counselors.” *The Journal of the American Medical Association* 278,15 (1997): 1217–20.

Discusses recommendations made by the National Society of Genetic Counselors concerning predisposition genetic testing for late onset diseases.

Merz, J. F. “Disease Gene Patents: Overcoming Unethical Constraints on Clinical Laboratory Medicine.” *Clinical Chemistry* 45,3 (1999): 324–30.

Deals with the threat that disease gene patents pose for physicians in providing medical care to their patients. Outlines the problems created by the monopolization of disease gene tests by a small number of providers and recommends amendment of patent law.

Mitchie, S., and T. M. Marteau. “Predictive Genetic Testing in Children: the Need for Psychological Research.” *British Journal of Health Psychology* 1 (1996), pp. 3–14.

Deals with the special problems associated with genetic testing of children.

Modell, B., et al. *Community Genetics Services in Europe: Report on a Survey*. WHO Regional Publications, European Series, No. 38, 1991.

A comprehensive account of genetic disease in Europe and the technologies and services available for treatment and prevention.

Murray, Thomas H. *Catch-22 and Genetic Discrimination* [available on the University of Pennsylvania Bioethics website: <http://www.med.upenn.edu/bioethics/ajem/ajem96/catch22.html>], 1996.

Examines the problem of discrimination in genetic testing for late onset diseases.

———. "Genetics and the Moral Mission of Health Insurance." *Hastings Center Report* 22 (1992), pp. 12–17.

While insurance companies already screen potential customers through physical exams, some fear that genetic screening will also be required either through direct testing or the disclosing of previously taken tests.

National Advisory Council for Human Genome Research. "Statement on Use of DNA Testing for Presymptomatic Identification of Cancer Risk." *Journal of the American Medical Association* 271 (1994), p. 785.

National Cancer Institute. *Understanding Gene Testing*. Publication No. 96-3905. Bethesda, MD: National Institutes of Health, 1995.

Clarifies gene testing in the context of cancer disease.

National Consultative Ethics Committee for the Life and Health Sciences. *Genetics and Medicine: From Prediction to Prevention.* Paris: Organisation for Economic Co-operation and Development, 1995.

Recognizes the lack of general knowledge of genetics and that the immediate practical solution to this is elusive. Calls for vigilance to ensure that information is being portrayed in media reports in a balanced and non-sensational fashion, and recommends genetic education starting at the secondary school level.

National Society of Genetic Counselors. *Resolutions: Prenatal and Childhood Testing for Adult Onset Disorders* [available on the Internet at <http://www.nsgc.org/ethicsResolutions.html>], 1995.

———. *Position Statements: Genetic Screening* [available on the Internet at <http://www.nsgc.org/ethicsPosition_Statements.html>].

Nuffield Council on Bioethics. *Genetic Screening: Ethical Implications*. 3rd ed. London, 1996.

Draws on experience of screening for diseases such as cystic fibrosis and sickle cell anemia. Examines issues such as consent, counselling, confidentiality and the possible use of genetic information by insurers or employers. Calls for the establishment of a central coordinating body to monitor genetic screening programs. The conclusions of the report have been widely endorsed. In 1996, the British government established two bodies to contribute to the regulation of genetics: the Advisory Committee on Genetic Testing and the Human Genetics Advisory Commission.

Oscapella, Eugene. *Genetics, Privacy and Discrimination*. A survey prepared for the International Working Group on Ethics and Public Confidence in Biotechnology, 1999.

Provides a cursory analysis of the relevant science relating to genetics and privacy, and an overview of the legislative schemes in Canada dealing with privacy and discrimination generally. Explores a broad range of genetic privacy and discrimination issues. Makes recommendations for specific action to protect genetic privacy and prevent discrimination.

Post, S. G. "Future Scenarios for the Prevention and Delay of Alzheimer Disease Onset in High Risk Groups: An Ethical Perspective." *American Journal of Preventive Medicine* 16,2 (1999): 105–10.

Discusses the two main goals of genetic testing of asymptomatic persons at high risk for Alzheimer disease: delaying onset of the disease through pharmacologic and lifestyle interventions and slowing the onset of moderate or advanced stages. A variety of ethical issues are considered.

Post, S. G., et al. "The Clinical Introduction of Genetic Testing for Alzheimer Disease. An Ethical Perspective." *The Journal of the American Medical Association* 277,10 (1997): 832–36.

Reports on the findings of a consensus group with participants in the areas of genetics, counselling, ethics and public policy who came together to address arguments for and against clinical genetic testing.

Rawbone, R. G.. "Future Impact of Genetic Screening in Occupational and Environmental Medicine." *Occupational and Environmental Medicine* 56,11 (1999): 721–24.

Considers a range of ethical issues with which the occupational health professional may be confronted as genetic technology advances.

Relkin, N. R., et al. "The National Institute on Aging/Alzheimer's Association Recommendations on the Application of Apolipoprotein E Genotyping to Alzheimer's Disease." *Annals of the New York Academy of Science* 16, 802 (1996): 149–76.

In a conference held in Chicago in October 1995, a working group of the National Institute of Aging and the Alzheimer's Association drafted consensus recommendations on research and clinical applications of APOE genetic susceptibility testing for Alzheimer disease.

Resnik, David B. "The Morality of Human Gene Patents." *Kennedy Institute of Ethics Journal* 7 (1997), pp. 43–61.

This author argues that many of the most pressing moral questions arising from patents on living material (particularly human genetic material or cell lines) involve important but nonetheless straightforward policy analysis.

Rhodes, Rosamond. "Genetic Links, Family Ties, and Social Bonds: Rights and Responsibilities in the Face of Genetic Knowledge." *Journal of Medicine and Philosophy* 23 (1998), pp. 10–30.

Addresses genetic privacy issues and the family.

Rosenthal, Thomas C., and Stirling M. Puck. "Screening for Genetic Risk of Breast Cancer." *American Family Physician* January 1, 1999.

Considers the difficulties and uncertainties surrounding genetic testing for breast cancer.

Roses, A. D. “Genetic Testing for Alzheimer Disease: Practical and Ethical Issues.” *Archives of Neurology* 54,10 (1997): 1126–29.

The utility of APOE genotyping is reviewed and recommendations for early use in diagnosis are explained. The ethical, social, actuarial and legal problems currently associated with genetic testing are discussed.

Rothenberg, Karen H. “Genetic Information and Health Insurance: State Legislative Approaches.” *The Journal of Law, Medicine and Ethics* 23 (1995), pp. 312–19.

Summarizes and analyzes U. S. state legislation on genetic information and health insurance. Highlights the major policy considerations that must be addressed in order to reach consensus on future strategies.

Rothstein, Mark A. “Discrimination Based on Genetic Information.” *Jurimetrics Journal* 33 (1992), pp. 13–18.

Discusses potential areas of genetic discrimination. Suggests that escalating healthcare costs and the principle of non-discrimination in employment appear to be on a collision course, and the Human Genome Project will accelerate the collision.

———. “Genetic Discrimination in Employment and the *Americans with Disabilities Act*.” *Houston Law Review* 29,1 (1992): 23–84.

Analyzes genetic discrimination in employment under the Americans with Disabilities Act. Specifically, it considers the coverage of certain genetic conditions under the Act, the restrictions on genetic testing by employers, the access to genetic information by employers, the legitimate uses of genetic information by employers, and the relationship between genetic information and employer-sponsored health insurance.

Sandberg, P. “Genetic Information and Life Insurance: A Proposal for an Ethical European Policy.” *Social Science and Medicine* 40, 11 (1995): 1549–59.

Searches for an ethical and feasible European policy for the use of genetic information in life insurance.

Schrag, Deborah, et al. “Decision Analysis: Effects of Prophylactic Mastectomy and Oophorectomy on Life Expectancy Among Women with BRCA1 and BRCA2 Mutations.” *New England Journal of Medicine* 336 (1997), pp. 1465–71.

Questions the value of genetic testing for susceptibility to breast cancer that will occur later in life. Healy, Bernardine. “BRCA Genes: Bookmaking, Fortunetelling, and Medical Care” is an accompanying editorial on the value of genetic testing for late-onset breast cancer.

Smith, George P.“Accessing Genomic Information or Safeguarding Genetic Privacy.” *Journal of Law and Health* 9 (1994–95), pp. 121–34.

Addresses the positive impact of accessing genomic information for promoting the social good. “Biological determinism” is necessary for transnational survival in the 21st century.

Smith, Ken R., and Robert T. Croyle. "Attitudes Toward Genetic Testing for Colon Cancer Risk." *American Journal of Public Health* 85,10 (1995): 1435–39.

Examines public interest regarding genetic testing for colon cancer susceptibility. Individuals with higher income and with a perceived risk of getting colon cancer were the most interested in testing.

Stranc, Leonie, and Jane Evans. *Issues Relating to the Implementation of Genetic Screening Programs*. Submitted to the Medical, Ethical, Legal and Social Issues Advisory Committee of the Canadian Genome Analysis and Technology Programme. Winnipeg: University of Manitoba, 1996.

Focusses primarily on the disease and test-specific issues surrounding the implementation of genetic screening. Examines genetic screening in the context of the classic World Health Organization (1968) guidelines, and reflects upon whether these are still germane in the context of genetic screening. The authors' conclude that the guidelines continue to be germane but need to be supplemented. Also, major areas of concern regarding genetic screening are raised, one dealing with the lack of efficacious treatments for the diseases for which screening can be done.

Tibben, Al, et al. "Psychological Effects of Presymptomatic DNA Testing for Huntington's Disease in the Dutch Program." *Psychosomatic Medicine* 56,6 (1994): 526–32.

Assesses the six-month follow-up effects of presymptomatic DNA testing for Huntington disease. Short-term positive effects on both carriers and non-carriers of the Huntington gene are noted and questions are raised about how foreknowledge will affect carriers as they approach the impending onset of the disease.

Troy, Edwin S. Flores. "The Genetic Privacy Act: An Analysis of Privacy and Research Concerns." *The Journal of Law, Medicine and Ethics* 25 (1997), pp. 256–72.

Reviews the Genetic Privacy Act and its commentary as they relate to the issue of an individual's right to privacy to genetic information, and the effects the Act may have on scientific research.

Vineis, P. "Ethical Issues in Genetic Screening for Cancer." *Annals of Oncology: Official Journal of the European Society for Medical Oncology* 8,10 (1997): 945–49.

Addresses conflict situations that emerge from genetic screening: for example, between respect for autonomy — the right not to know — and responsibility toward future generations — the duty to know for the sake of one' descendants. Considers some of the "hazards" of genetic testing for late onset diseases.

Vineis, P., and P. A. Schulte. "Scientific and Ethical Aspects of Genetic Screening of Workers for Cancer Risk: The Case of the Nacetyltransferase Phenotype." *Journal of Clinical Epidemiology* 48,2 (1995): 189–97.

Addresses scientific and ethical issues involved in the use of genetic screening techniques that intend to identify individuals that have more than average susceptibility to develop cancer from workplace chemical exposures. The case considered is the genetic polymorphism for N-acetyltransferase activity and the risk of bladder cancer in workers exposed to carcinogenic arylamines. The authors assert that genetic screening of workers for susceptibility to cancer is an ethically unacceptable and premature application of the science.

Wachbroit, Robert. "The Question Not Asked: The Challenge of Pleiotropic Genetic Tests." *Kennedy Institute of Ethics Journal* 8,2 (1998): 131–44.

Examines a moral dilemma created by one sort of pleiotropic testing, APOE genotyping, which can yield information about the risk of two conditions: coronary heart disease and Alzheimer disease. Explores how much providers should disclose to patients about pleiotropic test results and whether patients are obligated to know as much about their genetic condition as possible.

Wagner, T. M., and R. Ahner. "Prenatal Testing for Late-Onset Diseases Such as Mutations in the Breast Cancer Gene 1 (BRCA1): Just a Choice or a Step in the Wrong Direction?" *Human Reproduction* (England) 13,5 (1996): 1125–26.

Considers some ethical issues concerning prenatal testing for late onset diseases.

Weber, Barbara L. "The Crystal Ball: Genetic Testing for Inherited Susceptibility for Breast Cancer." *Ethics and Genetics Papers* [available on the Internet at <http://www.med.upenn.edu/bioethics/ajem/ajem96/crystal96.html>], 1996.

Discusses major issues raised by predictive testing for adult onset disorders. In particular, the issues of informed consent, privacy and long-term support and medical care.

Wertz, Dorothy C., and P. R. Reilly. "Laboratory Policies and Practices for the Genetic Testing of Children: A Survey of the Helix Network." *American Journal of Human Genetics* 61,5 (1997): 1163–68.

Highlights the results of a survey taken of DNA diagnostic laboratories in the U.S. The specific focus of the survey is related to the testing of children for late onset disease. The authors make recommendations concerning laboratory policies in regard to genetic testing of children.

Wiggins, Sandi et al. "The Psychological Consequences of Predictive Testing for Huntington's Disease." *New England Journal of Medicine* 327 (1992), pp. 1401–5.

Addresses psychological consequences of genetic testing for Huntington disease.

Wilson, J. M., and G. Jungner. *Principles and Practice of Screening for Disease*. Geneva: World Health Organization, 1968.

The authors of the World Health Organization genetic screening guidelines.

Wolf, Susan. "Beyond 'Genetic Discrimination': Toward the Broader Harm of Geneticism." *The Journal of Law, Medicine and Ethics* 23 (1995), pp. 345–53.

Argues that current U. S. state statutes and federal bills prohibiting genetic discrimination by health insurers demonstrate the inadequacy of an anti-discrimination approach to genetics. Considers why anti-discrimination analysis fails, and tries to get at the deeper underlying harm of discrimination in seeking alternatives.

Working Party of the Clinical Genetics Society (U.K.). "The Genetic Testing of Children." *Journal of Medical Genetics* 31 (1994), pp. 785–97.

Deals with the special problems of utilizing genetic testing for children.

Endnotes

1 T. Schrecker, et al., *Biotechnology, Ethics and Government*, p. 69.

2 *The New Shorter Oxford English Dictionary* (Oxford: Clarendon Press, 1993).

3 K. R. Melchin, "The Challenges of Technological Society," p. 123.

4 A. C. Upton, "Foreword," p. xi.

5 W. Gilbert, "A Vision of the Grail," p. 84.

6 D. J. Roy, et al., *Bioethics in Canada*, p. 450. Along this line of critique, see also the series of essays in C. F. Cranor (ed.), *Are Genes Us?*

7 Matt Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (New York: Harper Collins Publishers, 1999), p. 61.

8 N. Wexler, "Clairvoyance and Caution," p. 211.

9 These definitions are taken from four sources: N. A. Holtzman, and M. S. Watson (eds.), *Promoting Safe and Effective Genetic Testing in the United States*, pp. 6–7; Advisory Committee on Genetic Testing (United Kingdom), *Report on Genetic Testing for Late Onset Disorders*, pp. 8–9; "Proposed Genetic Screening Programmes: Proposed Recommendations of the European Society of Human Genetics," *European Journal of Human Genetics* 8 (2000), pp. 998–1000; and Australian National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: An Information Paper*, (Canberra: Commonwealth of Australia, 2000), pp. 63–71.

10 This is evidenced by the massive amount of study being done in Europe and North America in terms of legislation, policy statements and professional guidelines addressing either directly or indirectly the issue of genetic testing for late onset diseases.

11 "Population Genetic Screening Programmes," pp. 998–1000.

12 T. Lemmens, and P. Bahamin, *Genetics in Life,* p. 169.

13 T. H. Murray, "Assessing Genetic Technologies," p. 574.

14 Dorothy Nelkin, and M. Susan Lindee, *The DNA Mystique: The Gene as a Cultural Icon* (New York: W. H. Freeman and Company, 1995), p. 165.

15 N. R. Relkin, et al., "The National Institute on Aging/Alzheimer's Association Recommendations," p. 154.

16 Ibid., p. 155.

[17] A public opinion survey conducted by the March of Dimes Birth Defects Foundation in 1992 reported a significant lack of awareness among respondents of the social consequences of genetic testing. The survey "found that most Americans believe genetic information is public property and that those with a right to information about a person's genetic characteristics include not only those family members who could be immediately affected, but insurers and employers as well." Nelkin and Lindee, pp. 167–8. The report on the survey is by Louis Harris and Associates, Genetic Testing and Gene Therapy: National Survey Findings (White Plains, NY: March of Dimes Birth Defects Foundation, September 1992).

[18] *Human Genetics — Learning for the Millennium and Beyond*, Proceedings of the Human Genetic Advisory Commission's First National Conference at the Royal Society, October 16, 1998 [available at <http://www.dti.gov.uk/hgac/papers/papere1.htm>; website accessed March 2000].

[19] Available at <http://www.coe.fr/cm/ta/rec/1992/92r3.htm>; website accessed March 2000.

[20] Available at <http://who.org/ncd/hgn/hgnethic.htm>; website accessed March 2000.

[21] Available at <http://www.cdc.gov/genetics/publications/strategic.htm>, p. 18; website accessed March 2000.

[22] *Human Genetics — Learning for the Millennium and Beyond*.

[23] K. R. Smith, and R. T. Croyle, "Attitudes Toward Genetic Testing," p. 1437.

[24] Ibid.

[25] R. Hubbard, and R. C. Lewontin, "Pitfalls of Genetic Testing," pp. 1192–93. Similar criticism can be found in B. L. Weber, and Lemmens and Bahamin.

[26] Holtzman and Watson (eds.), p. 10. The Task Force was created by the National Institutes of Health-Department of Energy Working Group on the Ethical, Legal and Social Implications of Human Genome Research.

[27] National Consultative Ethics Committee, *Genetics and Medicine*; Ruth Chadwick, et al., *Ethical Implications of Human Genome Analysis for Clinical Practice in Medical Genetics with Special Reference to Genetic Counselling*, a report to the Commission of the European Communities (Cardiff: Centre for Applied Ethics, 1993).

[28] The issues of confidentiality and discrimination are dealt with in separate sections.

[29] Advisory Committee on Genetic Testing, *Report on Genetic Testing,* p. 17.

[30] Ibid., p. 18.

[31] Ibid.

[32] T. A. Caulfield, *The Commercialization of Human Genetics*, p. 18.

[33] Holtzman and Watson (eds.), p. 13.

[34] Ibid.

[35] Ibid.

[36] P. J. Bridge, *Genetic Testing for Predispositions*, p. 41.

[37] Advisory Committee on Genetic Testing, p. 26.

[38] Holtzman and Watson (eds.), appendix 4.

[39] Ibid., p. 61.

[40] Hubbard and Lewontin.

[41] Advisory Committee on Genetic Testing, pp. 21–22 and Holtzman and Watson (eds.), p. 8. In the United Kingdom, obtaining written consent is already an established practice when testing for Huntington disease and most other serious late onset diseases.

[42] Advisory Committee on Genetic Testing, p. 23 and Holtzman and Watson (eds.), p. 8.

[43] See, for example, "Presymptomatic and Susceptibility Testing," *Proposed International Guidelines*, World Health Organization, and "The European Perspective," *Ethical Implications of Human Genome Analysis for Clinical Practice in Medical Genetics*, a report to the Commission of the European Communities (Paris: Organization for Economic Cooperation and Development, 1996). This text looks specifically at social values and ethical traditions in relation to genetic counselling in Denmark, Germany, Italy, the Netherlands and the United Kingdom.

[44] B. Bowles Biesecker, "Future Directions," p. 145.

[45] Barbara Bowles Biesecker is a genetic counsellor and co-director of the Genetic Counseling Research and Training Program in the Medical Genetics Branch of the National Human Genome Research Institute at the National Institutes of Health.

[46] A. Clarke, "The Genetic Testing of Children," *The Ethics of Genetic Screening*, Ruth Chadwick, et al., eds. (Dordrecht: Kluwer Academic Publishers, 1999), pp. 231–47; P. S. Harper and A. Clarke, "Should We Test Children for 'Adult' Genetic Diseases?" *Lancet* 335 (1990), pp. 1205–6; and Institute of Medicine, *Assessing Genetic Risks*, L. B. Andrews et al., eds.

[47] D. C. Wertz, and P. R. Reilly, "Laboratory Policies and Practices," p. 1163.

[48] Advisory Committee on Genetic Testing, p. 27 and Holtzman and Watson (eds.), p. 9.

[49] Wertz and Reilly, p. 1165.

50 Ibid., p. 1167.

51 See Caulfield, p. 13, where he refers to a survey of Canadian genetic professionals. “Fifty three percent of the surveyed professionals thought they should refer a patient outside of Canada if domestic law forbids a requested genetic service — a particularly potent statistic given the prohibitions proposed in Bill C-47.”

52 Advisory Committee on Genetic Testing, p. 28.

53 These are discussed in detail in the third section of this report.

54 Advisory Committee on Genetic Testing, p. 29.

55 Roy, et al., p. 169.

56 Ibid., pp. 187–89.

57 These categories are drawn from the Advisory Committee on Genetic Testing report *Advice to Research Ethics Committees: Points to Consider in Ethical Review of Medical Research Involving Genetic Testing*, released October 1998 and available at <http://www.doh.gov.uk/genetics/recrev3.htm>. This report was released following a request from research ethics committees for advice on research trials that included genetic testing. This thorough and comprehensive document provides questions for committees to use when evaluating these studies.

58 Advisory Committee on Genetic Testing, p. 30.

59 Ibid., pp. 3–6.

60 Holtzman and Watson (eds.), p. 10.

61 “In using the knowledge arising from research on APOE and its association with Alzheimer’s Disease, the interests of patients, future patients and family members should be paramount.” Relkin, et al., p. 161. This expresses well the consensus in Europe and North America concerning research studies that involve genetic testing for late onset diseases.

62 Ibid., p. 11.

63 Holtzman and Watson (eds.), p. 12.

64 These principles are taken from S. Burris and L. O. Gostin, “Genetic Screening from a Public Health Perspective: Some Lessons from the HIV Experience,” *Genetic Secrets*, M. A. Rothstein (ed.), pp. 141–45.

65 Hubbard and Lewontin, p. 1193.

66 Steven S. Coughlin, et al. “BRCA1 and BRCA2 Gene Mutations and Risk of Breast Cancer: Public Health Perspectives,” *American Journal of Preventative Medicine* 16,2 (1999): 96.

67 Ibid.

68 S. Le Bris and B. M. Knoppers, "International and Comparative Concepts of Privacy," Rothstein (ed.), pp. 418–19, identify four factors: technological advances and the shift from "cloistered" societies to mass culture, the development of the media and the subsequent conflict between privacy and the right to information, the evolving importance of one's "honour" and one's "image," and the emergence of pluralistic societies where "the private domain has gradually been extended and state control has gradually diminished."

69 Ibid., p. 419.

70 "The Privacy Commission of Canada has stated that no surveillance technology is more threatening to privacy than that designed to unlock the information contained in human genes." Roy, et al., p. 453.

71 The issue of privacy protection to non-existing future generations in the context of genetic testing is discussed in J. Sandor, "Genetic Testing, Genetic Screening and Privacy," Chadwick et al. (eds.), pp. 181–90.

72 Roy, et al., p. 453.

73 For example, *Genetic Secrets*, Rothstein (ed.), deals with the ethical, practical and legal ramifications of genetic testing. From the U.S. perspective, important articles dealing with this are L. O. Gostin, "Genetic Privacy," pp. 320–30, and G. J. Annas et al., "Drafting the Genetic Privacy Act" pp. 360–66. Both articles are important because they attempt to wrestle with the unique problems genetic information presents. As Annas et al., p. 365, states, "The gene has become more than a piece of information; it has become 'a cultural icon, a symbol, almost a magical force.' To the extent that we accord special status to our genes and what they reveal, genetic information is uniquely powerful and uniquely personal, and thus merits unique privacy protection." Also, G. P. Smith, "Accessing Genomic Information," pp. 121–34. From the Canadian perspective, there is E. Oscapella, *Genetics, Privacy and Discrimination*, and W. F. Flanagan, "Genetic Data and Medical Confidentiality," pp. 269–88.

74 Holtzman and Watson (eds.), p. 9.

75 Ibid.

76 See, for example, P. M. Schwartz, "European Data Protection Law and Medical Privacy" and Le Bris and Knoppers, Rothstein (ed.), pp. 392–417 and 418–48.

77 Schrecker, p. 69.

78 Roy, et al., p. 454.

79 D. Nelkin, and M. S. Lindee, *The DNA Mystique*, p. 167.

80 Lemmens and Bahamin, p. 25.

[81] An example of the link between genetic information, discrimination and racial prejudice occurred in 1970 when the American federal government offered free genetic testing for falciform anemia (a recessive hereditary disease found more frequently in the black population than other groups; it is not a late onset disease). Some tested positive as carriers and others developed the disease. The well-known boxer Joe Fraser encouraged black people to take the test. Eventually, some employers refused to hire people who carried the gene under the "scientific" pretext that such employees may faint on the job. The U.S. Air Force refused to employ blacks who were carriers of a gene with anemic characteristics. An airline company even dismissed some of its stewardesses for the same reason. The DuPont company excluded all black carriers of the gene. As well, many institutions, banks and industries confused the carriers of the gene with those who were affected with the disease. The project discontinued when the Black Panthers denounced the screening project as a planned ethnocide. J. P. Rogel, *La grande saga des gènes humains* (Paris: Lanctot Éditeur, 1999), pp. 95–97.

[82] Holtzman and Watson (eds.), p. 9.

[83] This is quoted in Lemmens and Bahamin, p. 25, footnote 95.

[84] Nelkin and Lindee, p. 168.

[85] Ibid., p. 174.

[86] Available on the Internet at <http://www.dti.gov.uk/hgac/papers/papers_g/g_01.htm>. This Commission is now closed and its responsibilities have passed to the Human Genetics Commission.

[87] Rothstein (ed.), p. 282.

[88] L. B. Andrews and A. S. Jaeger, "Confidentiality of Genetic Information in the Workplace," p. 77.

[89] Available at <http://www.dti.gov.uk/hgac/papers/papers_g/g_02.htm>; website accessed March 2000.

[90] Rothstein (ed.), p. 296.

[91] M. A. Rothstein, "Genetic Discrimination in Employment."

[92] P. Sandberg, "Genetic Information and Life Insurance," p. 1549. Sandberg is quoting the U.S. National Institutes of Health/Department of Energy Task Force on Genetic Information and Insurance.

[93] Lemmens and Bahamin, p. 5.

[94] Human Genetic Advisory Commission, *Second Annual Report 1999*, paragraph 2.1.

[95] As quoted in Sandberg, p. 1553.

[96] A quote from J. A. Lowden, Vice-President and Chief Medical Director of one of the major Canadian insurance companies in Lemmens and Bahamin, pp. 2–3. There are many examples of insurance companies indirectly refusing insurance in relation to genetic testing. For example, there are isolated cases in which insurance companies have refused coverage to some families with a history of Huntington disease, but have left the door open to coverage if the individuals receive negative genetic test results (Ibid., p. 3, footnote 10). The widow of a Chicoutimi man who died in a car accident was denied payment of her husband's life insurance because he had failed to report the positive test results for Steinerts dystrophy. The genetic screening had been part of a research study eight years prior to his application for life insurance. The Quebec Superior Court argued in favour of the insurance company because the man was guilty of false representation for answering negatively to the question asking whether he presented with any physical or mental anomalies. J. P. Rogel, *La grande saga*, pp. 166–69.

[97] Sandberg, p. 1557.

[98] Ibid., p. 1550.

[99] Ibid., p. 1552.

[100] Marcel J. Mélançon, and Richard Gagné, *Dépistage et diagnostic génétiques: Aspects cliniques, juridiques, éthiques et sociaux* (Laval: Les Presses de l'Université Laval, 1999), pp. 112–14.

[101] Robert J. Pokorski, "Insurance Underwriting in the Genetic Era," *American Journal of Human Genetics* 45 (1997), pp. 647.

[102] Sandberg, p. 1556.

[103] James F. Childress, "The Art of Technology Assessment," *On Moral Medicine: Theological Perspectives in Medical Ethics*, Stephen E. Lammers and Allen Verhey, eds. (Cambridge: William B. Eerdmans Publishing Company, 1998), p. 298.

[104] Edward Leroy Long, "Technology," *The Westminster Dictionary of Christian Ethics*, James F. Childress and John MacQuarrie, eds. (Philadelphia: The Westminster Press, 1986), p. 617.

[105] For example, T. H. Murray, "Assessing Genetic Technologies," pp. 573–82, and J. F. Childress, "The Art of Technology Assessment," S. E. Lammers and A. Verhey, eds., pp. 297–308.

[106] Neil A. Holtzman, "Promoting Safe and Effective Genetic Tests in the United States: Work of the Task Force on Genetic Testing," *Clinical Chemistry* 45,5 (1999): 734.

[107] Hubbard and Lewontin, p. 1193.

[108] As noted earlier in this report, commercial interests in the United States and Europe started marketing APOE genotyping before its usefulness had been established. Relkin, et al., p. 154.

[109] "For the vast majority of people affected by heart disease, cancer and the like, the origin is so complex that it's a gross oversimplification to think that screening for a predisposing gene will be predictive." Lemmens and Bahamin, p. 21. Also, Nelkin and Lindee, pp. 165–66, argue that "predisposition in the clinical sense is a statistical risk calculation, not a prediction. A person 'predisposed' to cancer, for example, may have biological qualities that heighten the odds that he or she will develop cancer, in the same way that driving many miles each day heightens one's odds of involvement in an automobile accident. But many variables influence whether a person will actually suffer from cancer. Terms such as "predisposed"or "at risk" are understood by scientists to mean that the individual is vulnerable to a disease that may *or may not* be expressed in the future."

[110] The important work of the U.S. Task Force on Genetic Testing in this regard is noted above. In addition, the Stanford Program in Genomics, Ethics, and Society has made several recommendations concerning the "criteria for judging the readiness of a test for introduction into routine clinical practice." L. M. McConnell, et al. "Genetic Testing and Alzheimer Disease," p. 3. The National Institute on Aging and the Alzheimer's Association have made recommendations. Also, the U.K. Advisory Committee on Genetic Testing highlights in its report the importance of scientific and clinical validity of the test. The problem of "premature implementation" has been noted in Caulfield, *The Commercialization of Human Genetics*, pp. 18–23. One of the recommendations from Bridge, p. 41, is "that uniform mechanisms are developed to evaluate when a test is suitable to offer."

[111] Advisory Committee on Genetic Testing, p. 10.

[112] Bridge, p. 4.

[113] Rogeer Hoedemaekers, "Genetic Screening and Testing," *The Ethics of Genetic Screening*, Ruth Chadwick, et al., eds (Dordrecht, Netherlands: Kluwer Academic Publishers, 1999), p. 209.

[114] An excellent example of this can be found in Viktor E. Frankl's book *Man's Search for Meaning* (New York: Washington Square Press, Simon and Schuster, 1963). Frankl's observations centre on his experience in a German concentration camp in the Second World War. What astonished him was that despite dehumanizing conditions, many prisoners sought meaning in their experience, and that this search gave them a heightened sense of the worthwhileness of existence despite the horrendous conditions in which they were living.

[115] Christopher Young, "Descent into Alzheimer's," *Maclean's*, March 13, 2000, pp. 30–31.

[116] An important article on privacy and its manifestations in various jurisdictions and its impact on policy formation is Le Bris and Knoppers, "International and Comparative Concepts of Privacy," Rothstein (ed.), pp. 418–48.

117 A brief comparative overview of international and national developments on the confidentiality of health information is given in Bartha Maria Knoppers, et al., *Opportunities-Barriers for Access to Use of Cancer Information for Surveillence Purposes*, (at press) 104ff.

118 There is tremendous awareness in North American and Europe of the importance of genetic education. This is clearly stated in the policy statements that are emerging from various study groups and task forces. One Quebec author stresses the importance of genetic information relating to hereditary diseases being offered in high schools, CEGEP's and universities. Gérard Tremblay, "Les parents face au dépistage génétique," *Dépistage et disgnostic génétiques,* M. J. Mélançon, and R. Gagné, eds., pp. 33–41.

119 For example, in Austria, Section IV of *Federal Law 510* states that "gene analysis in man for medical purposes may only be done at the request of a physician specialised in Human Genetics or by a physician specialised in the relevant clinical field for … verification of a predisposition for a disease, especially for predisposition of a possibly later onset hereditary disease …." Gertrud Hauser, "Genetic Screening, Information and Counselling in Austria," Chadwick et al., p. 39. In Belgium, "a total ban on the use of genetic testing to predict the future health status of applicants for (life) insurance was laid down in the *Law on Insurance Contracts*, which came into force in September 1992 …. A complete prohibition on communication of genetic data to insurers without any exception was laid down in the same *Law on Insurance Contracts*." Kris Dierickx, "The Belgian Perspective on Genetic Screening," Ibid., p. 51.

120 See, for example, the 1993 *Report to the Commission of the European Communities: Ethical Implications of Human Genome Analysis for Clinical Practice in Medical Genetics with Special Reference to Genetic Counselling*.

121 This issue touches very directly the whole debate around the testing of children for late onset diseases.

122 R. Hoedemaekers, "Genetic Screening in the Netherlands," *The Ethics of Genetic Screening*, R. Chadwick et al., eds., p. 115.

123 Ibid., p. 116.

124 One exception is the treatment given to geneticization in the Australian report by the National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: An Information Paper*. The report acknowledges the concern for "over concentration on research on genes and their health implications [leading] to neglect of the effects on human health of other factors, such as the physical, social and economic environments in which people live." It also notes that geneticization could threaten social solidarity. "An example of loss of solidarity in society would be the expectation that those with genetic susceptibilities, or at risk of having children with a genetic disorder, increasingly take financial responsibility for their own and their affected children's health care. It would be argued by those supporting such a policy that individuals have a duty to prevent illness in

themselves and not to have children with genetic disorders. They would also argue that if preventive measures are not taken, society should not have to provide the resources needed for care of those affected. Such attitudes would be an attack on the prevailing view that health care costs should be distributed across the whole community, and would challenge concepts of equality of respect for persons and sense of community," p. 14.

125 National Consultative Ethics Committee, p. 1.

126 Ibid., p. 2.

127 Ibid., p. 3.

128 Ibid.

129 Ibid., pp. 4–6.

130 Ibid., p. 6.

131 See, for example, N. Lenoir and B. Mathieu, *Les normes internationales de la bioéthique*, especially chapter IV, "Les norms universelles de la bioéthique," pp. 97–119; Council of Europe, "Convention for Protection of Human Rights," pp. 277–290; and N. Lenoir, "UNESCO, Genetics, and Human Rights," pp. 31–42.