Canadian Biotechnology Advisory Committee

Protecting Privacy in the Age of Genetic Information

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PREFACE

Advances in genetics are often hailed as the first step in finding a treatment or cure for serious diseases or conditions. Many announcements of such advances point out that it may still be years, if ever, before any therapy will be available to patients. Despite this reality, there is a sense that these advances in genetics will ultimately result in many health benefits. This perception may well encourage people to participate in genetic research as a way of bringing those projected benefits closer. While the willingness of individuals to participate in genetic research is laudable, concerns have been raised about possible violations of the privacy rights of these individuals. Should genetic information be treated like other medical information, or does it have characteristics that make it unique? Should it be protected in some circumstances, but made available in others? The inappropriate release or use of genetic information could lead to genetic discrimination, which might take the form of rejection for employment, loss of credit, insurance, eligibility for pensions, or even discriminatory treatment in the application of government social policy. The unwanted sharing of genetic information could also cause rifts in families between members who want to know if they are at risk for a certain genetic disease and other members who wish not to know. Above all, the promise of genetic research to improved health may be jeopardized unless privacy and discrimination issues are addressed; unless people are satisfied that their information will be appropriately protected, they will not participate.

From the time it was formed, the Canadian Biotechnology Advisory Committee (CBAC) recognized privacy issues related to genetic information as an area of public concern. In 2000, CBAC commissioned two papers, one to look at the pace of genetic research and its implications and the other to consider the potential for individuals to be discriminated against on the basis of their genetic information. Two years later, the Committee asked a team of legal experts, ethicists and researchers to probe deeper in order to examine the legal, ethical and social implications of large-scale population genetic research and information storage. Drafts of these papers were presented at a half-day workshop session at Genome Canada's first annual GE3LS Symposium in February 2003. Once the papers were finalized, a synthesis was prepared.

With this publication, consisting of the two background papers and the synthesis paper, CBAC provides background and some possible answers, informed opinion and recommendations to guide legislators and others as they grapple with these issues. The first paper explores the possible uses of genetic information, while the second examines some of the legal ramifications of those uses. The third paper revised issues related to the establishment of large collections of genetic information ("biobanks") intended to be representive of a general population. This paper summarizes legal and social issues arising from research using these biobanks and describes the views of researchers, health professionals and the general public concerning biobanks. It also proposes a framework for ensuring that the establishment of population biobanks in Canada provides both benefits for Canadian society and privacy protection for the individuals whose genetic information is included in them.

INTRODUCTION

Almost daily, we read of advances in science centred on the unravelling of the map of the human genome. Words such as "miracle" are used freely to describe these discoveries. Indeed, the possibilities of genetic research are profound and the potential benefits to human health are enormous. But to realize these benefits, researchers will need access to the DNA of thousands of people. How researchers obtain these samples and what they do with them are critical to whether these potential benefits can be achieved.

Public opinion research has consistently revealed that Canadians feel there is something very personal and very special about their genetic information. It is not just medical or health information, it is information about one's very identity. We would do well, therefore, to be cautious in the uses we sanction for genetic information.

While Canadians are open to participation in genetic research, they guard their privacy jealously. As the CBAC's own research has revealed, they also become increasingly uneasy once they learn about the potential medical and non-medical uses to which their information could be put.

There are few who dispute the propriety of using genetic information to diagnose and treat illness, identify missing persons or solve crimes. But, as Trudo Lemmens and Lisa Austin point out in their paper entitled "Of Volume, Depth and Speed: The Challenges of Genetic Information," the potential uses of genetic information go far beyond what many people have conceived. What is not widely known by Canadians is that there is a movement to use genetic information for purposes far removed from human health. These new uses for genetic information raise a whole new set of questions and concerns related to personal privacy. These two authors warn that, if left unchecked, actual intrusions into personal privacy could become much greater than ever anticipated, propelled by the very technology that has expanded the horizons of genetic research. From their perspective, the computer, coupled with the volume of information that can already be extracted from a single genetic sample and the speed of testing, threatens to compromise our privacy as never before. They conclude

that new regulatory measures must be developed – or existing measures adapted – to protect both individuals and society as a whole.

Some of the most spectacular advances in medicine in recent decades have been obtained through DNA analysis. Indeed, part of the "promise" of genetic research is its well-established role in the early detection and, in some cases, treatment of disease. Genetic research is enabling us to unravel the mysteries of Huntington disease, breast cancer, Tay Sachs disease and some mental illnesses.

This has assisted people to make life choices, take preventive action if possible and increase monitoring to be able to identify the disease at an early stage. At the same time, fears are raised that genetic profiling could also result in their exclusion from insurance, discrimination in employment, stigmatization as members of "diseased" families, and emotional suffering – whether they ever actually come down with the disease. The challenge, therefore, is to find a way to harness this new knowledge while limiting any negative consequences.

As Eugene Oscapella argues in his paper "Genetics, Privacy and Discrimination," the privacy issue is one that genetic researchers and policy makers ignore at their peril:

> *The possible use of personal genetic information against individuals may justifiably stifle acceptance of further genetic inquiry. Failure to protect privacy and prevent discrimination therefore risks greatly diminishing the potential for genetics to improve health care.*[1]

One of the more troubling issues raised by Oscapella is what he describes as the "rapid advance of genetic science." He notes that existing laws governing personal health information also, sometimes explicitly, often protect genetic information used for health purposes. However, these protections are, at best, a patchwork and are "incomplete," given many of the unanticipated issues that have already arisen from genetic science. Therein lies one of the challenges for policymakers.

For an examination of the ethical, legal and social issues associated with the growth of large-scale genetic research, the Canadian Biotechnology Advisory Committee commissioned four

[1] E. Oscapella, "Genetics, Privacy and Discrimination." (2000), p.3

papers on various aspects of population biobanks, Mylène Deschênes and Geneviève Cardinal surveyed a variety of national approaches to the development of genetic biobanks. Patricia Kosseim produced an overview of relevant Canadian legislation at all levels of government. Michael Yeo studied the privacy issue as it relates to storage, retrieval and use of genetic information. Edna F. Einseidel examined public and professional views of biobanking. Drafts of these papers were presented at Genome Canada's first annual GE3LS Symposium in February 2003. After the papers were completed, Lorraine Sheremeta then synthesized and expanded their research. When considered together, their findings are likely to heighten the concerns of Canadians.

So far, there is much more promise to genetic research and biobanking than there are actual results. Because of this, there is a temptation to dismiss or delay consideration of some of these authors' more pointed questions. Yet, as Michael Yeo observes in his paper "Research and Biobanking: The Conflict Between Privacy and Access Made Explicit," the tension between our thirst for knowledge and commercialization of biotechnological breakthroughs and the rights and values we share as Canadians is real and increasing.

One of the strongest messages to come from these authors is that the future of genetic research itself rests squarely on the shoulders of public confidence. As Sheremeta underlines in her synthesis paper, "The governance of biobanks is critically important because of the role it plays in ensuring accountability and in building and maintaining public trust."[2] She also cautions that a failure to apply the highest scientific, legal and ethical standards will inevitably undermine public trust and confidence in scientific development and the products of such research.

Biobanking has been greatly facilitated by computer technology. As is the case with the banks where we keep our money, much of the 'wealth' of biobanking is stored in electronic format rather than as actual DNA samples. This has made data storage much easier and has opened up whole new vistas of research. It has also re-ignited a long-simmering debate over a range of issues centred on privacy and confidentiality, recruitment of DNA donors and informed consent, discrimination, commercialization and governance.

These are precisely the issues that the Canadian Biotechnology Advisory Committee addresses with this report as part of its mission to provide advice to policymakers.

[2] Sheremeta, p. 52

OF VOLUME, DEPTH AND SPEED: THE CHALLENGES OF GENETIC INFORMATION

Document Prepared for the Canadian Biotechnology Advisory Committee

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February 2001

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1. INTRODUCTION

The futuristic movie GATTACA pictures the struggle of a 'genetic proletarian,'[1] born out of a stubborn mother who refused to abort her genetically inferior fetus, towards the fulfilment of his life-dream of becoming a cosmonaut. In order to do so, he has to cheat the genetically monitored GATTACA society that performs regular DNA testing to systematically screen out people of his kind from any reasonable job and from insurance coverage. One of the scenes of the movie portrays the activities of a local "gene shop." Clients of this shop have a mouth swab to recuperate DNA traces of the person they just dated and kissed. The gene shop conducts a computerized DNA analysis on the spot, providing a summary genetic portrait of the potential partner they just 'caught.' On a one-page summary, clients get basic information about the person's behavioral traits, life-expectancy and potential progeny.

Obviously, a simple mouth swab and DNA analysis based on saliva currently cannot give us such detailed information and certainly not in a time span of three minutes, as suggested in the movie. Also, several scientific hurdles have to be overcome before we will be able to conduct simultaneous, and affordable, tests for a variety of conditions and traits. But even though the gene-shop scene is very much a caricature of genetic testing, it provides a strong metaphor both for what type of issues are likely to be raised by genetic testing and for why we have to be concerned about the social consequences of unbridled use by third parties. Research into the development of DNA chip and microarray technology is already taking place and in the near future will likely allow us to scan entire genes for the detection of a variety of mutations. Genetic tools will become faster, more efficient, and cheaper.[2]

1 The term is taken from R.C. Dreyfuss & D. Nelkin "The Jurisprudence of Genetics" (1992) 45 Vanderbilt L. Rev. 313 at 318

2 For a recent overview of new genetic technology and its impact on medicine, see K.M. Kurian, C.J. Watson & A.H. Wyllie, "DNA chip technology [editorial]" (1999) 187(3) Journal of Pathology 267, 1999; see also S.J. Watson & H. "Gene chips and arrays revealed: a primer on their power and their uses." (1999) 45(5) Biological Psychiatry 533; for a view on the ethical implications of the introduction of this technology, see W. Henn, "Genetic screening with the DNA chip: a new Pandora's box?" (1999) 25(2) Journal of Medical Ethics 200.

In this paper, we will argue that the combination of the following three elements constitute the primary reason why we have to develop appropriate regulatory measures or adapt existing ones:

1) the volume of information that can be extracted from one sample;

2) the speed of testing; and

3) its link with computer technology.

These are the main reasons, we argue, why genetic testing, if inappropriately used, can have detrimental social consequences. Other characteristics have been identified as making genetic information 'unique', but we will argue that many other types of health information share these characteristics. However, when combined with the three factors we identify, many of our traditional concerns regarding health information are augmented. In other words, the concerns raised by the advent of genetic testing are related more to what one can call an amplification of existing concerns about the use of health information than to the specificity of genetics. It is a matter of degree, or depth, more than a matter of newness. But even if these concerns are not in themselves new, the new contexts in which they are raised may require different types of responses, or additional responses, than those pertaining to more traditional health information.

This paper aims at identifying the relative specificity of genetic information and analyzing the arguments invoked to support specific regulation and legislation that singles out genetics.

2. WHAT IS GENETIC INFORMATION?

It is difficult to analyze the use of genetic information in a comprehensive manner without treating genetic information as a one-dimensional concept. However, one always has to keep in mind that genetic information can be obtained in a variety of ways and can refer to very different forms of health information. The genetic information referred to in this paper is generally the information resulting from genetic research undertaken with new genetic technology developed in the last decades and that has led to the identification of specific associations between genes and genetic diseases and traits. It is this relatively new form of genetic information that much of the debate about the potential negative social impact of genetic testing focuses on. But the term genetic information also includes family history of disease, information from chromosomal testing and data gathered from twin studies, for example, all which have been used in research and in health care for the most part of the last century without receiving the same attention. In the debate over what constitutes genetic information, some even point out that all health information is to some extent genetic.[3]

Any type of regulation or legislation developed in the context of genetics will have to be attentive to the problem of defining what constitutes genetic information and how one can distinguish in fact, and as a matter of principle, the different types of genetic data. In order to highlight this problem, we will enumerate here some of the major differences between types of genetic information arising from the type of testing, the purpose and manner of collection of the information or sample, and the method of storage.

2.1 DNA Testing

In most current discussions, genetic information is understood as information resulting from the analysis of an individual's DNA. Startling developments in molecular genetics and DNA technology (closely linked also to developments in computer technology) over the last decades are directly responsible for what has been termed ‘the genetic revolution’. When people talk about ‘genetic information,’ they are most likely thinking of information derived from the use of this new technology. Through the use of a variety of techniques such as

[3] See *e.g.*, J.S. Alper & J. Beckwith, “Distinguishing Genetic From Nongenetic Medical Tests: Some Implications For Antidiscrimination Legislation” (1998) 4 Science and Engineering Ethics 141 at 148.

electrophoresis, somatic cell hybridization, cytogenetic mapping, multiplexing, and radiation-induced breakage of chromosomes, scientists have been able to make physical maps of the human genome. The physical maps portray the position, size, order and numbering of base pairs in the different genes. Comparison of the maps of different people allows researchers to find specific mutations associated with genetic conditions or traits. Even when the mutation directly related to a genetic condition has not been identified, DNA techniques can be used to find markers for the disease. Markers are characteristic DNA sequences that enable scientists to determine whether a mutation present in that DNA region has been inherited or not. A variety of tests have been developed on the basis of these techniques. Currently available DNA-based genetic tests include tests for: Amyotrophic lateral sclerosis (Lou Gehrig's disease), Alzheimer disease, ataxia telangiectasia, inherited breast and ovarian cancer, Cystic Fibrosis, Duchenne muscular dystrophy, fragile X syndrome, Huntington's disease, myotonic dystrophy, sickle cell disease, thalassemia, Tay-Sachs disease and many other conditions.[4]

2.2 Indirect Genetic Testing

Although some of the most spectacular advances in medicine have been obtained by DNA analysis, other forms of testing can clearly be identified as 'genetic tests.'[5] The identification of phenotypic characteristics associated with genetic conditions such as cleft palate or Spina Bifida, for example, is a form of genetic testing. Testing can also occur at the chromosomal level. Chromosomal abnormalities can be detected, for example, through amniocentesis. Other forms of 'genetic tests' involve the testing of urine, blood or other body fluids to discover abnormal metabolite levels that are indicators of genetic disorders such as phenylketonuria (by measurement of phenylalanine in blood) or Lesch-Nyhan disease (by identification of high urinary uric acid levels). Finally, genetic disorders can be detected through measuring proteins, which are the products of genes. Defective genes often lead to identifiable deficiencies in protein production. The observation of mutant proteins can be used as a measurement to determine the presence of a genetic condition such as Tay-Sachs.

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4 For a list of tests, see *e.g.* D.K. Casey, "What Can the New Gene Tests Tell us?" (1997, Summer) The Judges Journal 14 at 15.

5 An overview of various forms of genetic testing can be found in Science Council of Canada, *Genetics in Canadian Health Care* (Ottawa: Minister of Supply and Services, 1991), in particular at 37-42 [hereinafter *Genetics in Canadian Health Care*]; K.C. Glass, C. Weijer, T. Lemmens, R. Palmer and S.H. Shapiro, "Structuring the Review of Human Genetics Protocols, Part II: Diagnostic and Screening Studies." (1997) IRB, A Review of Human Subjects Research 19(3-4): 1 at 4; and Human Genetics Commission, *Whose Hands on Our Genes: A Discussion Document on the Storage Protection and Use of Personal Genetic Information* (London, s.d., s.l.), [accessed at http://www.hgc.gov.uk/] [hereinafter *Whose Hands on Our Genes*]

2.3 Family History

For a very long time, people have been aware of the fact that diseases are 'running in families' and have been involved in studying the familial character of diseases. Not only family physicians, but also interested third parties such as insurance companies have been aware of this and have been collecting information on people's family history of disease. Indeed, from time immemorial people have talked about people having a disease 'running in the family.' The history of behavioral genetics contains a remarkable example of a lay person's contribution to genetic research. The first association of a particular gene with a tendency to violence was established with crucial help from detailed records of a Dutch family's history of crime and violence, kept by one member of that family. Clearly, family history of disease is genetic information that can lead to the identification of 'at risk families', in which all members are identified at increased risk for developing certain conditions. For example, breast cancer, Huntington's disease, Tay Sachs, and some mental illnesses, have all been 'running in families' and people identified as members of these families have both benefited from knowing this (e.g. for making life choices, taking preventive action if possible, improved monitoring) as well as been harmed by it (e.g. being excluded from insurance, discriminated in employment, being stigmatized as members of diseased families, suffering emotionally).

2.4 Differentiation of Genetic Testing According to its Health Care Purpose and its Timing

Genetic testing can be differentiated according to why it is used in health care and at what stage.

Pre-natal diagnosis
This is genetic testing that is being conducted before birth to determine whether a foetus is affected by or at risk for having a genetic disorder.

New-born screening
New-born screening focuses on the identification of metabolic disorders in neonates, for which early treatment may be crucial to reduce the progression of the disease. New-born screening exists for a variety of conditions such as phenylketonuria, galactosemia and homocystinuria.

Pre-symptomatic testing

This is carried out on healthy individuals to determine whether they carry a genetic mutation that increases their likelihood of developing a genetic condition. It aims at determining people's future health risks, and generally does not relate to their present health status. The predictive character of the tests will vary according to the type of disorder tested for, but the term pre-symptomatic testing is generally used for more 'determinant,' late-onset genetic conditions. These are the traditional conditions in which a positive test result indicates a very high likelihood of future illness. A paradigm example is Huntington's disease, a dominant, single-gene disorder.[6]

Diagnostic genetic testing

In its strictest sense this form of testing aims at confirming a particular diagnosis through a genetic test. Lesch-Nyhan disease, for example, can be diagnosed by conducting an enzyme assay. Conducting a genetic test likely will become a standard part of many diagnoses. In the domain of mental health care, for example, there is an expectation that genetic research will promote more accurate diagnosis and better treatment targeted at subcategories of mental health disorders that are currently not clearly discernible because of the lack of precise clinical tools.[7]

It is important to note that genetic research and diagnostic genetic testing may impact on the typology of a disease. For example, new research indicates that some people who carry the cystic fibrosis gene may have none of the most severe expressions of the disease. [8] It shows that some mutant genes tend to be not associated with the traditional pulmonary disease of CF (i.e. early onset of progressive bronchiectasis). However, people having the mutant gene may suffer from related health problems also associated with cystic fibrosis such as pancreatitis and reproductive problems, in particular in the form of absence of *vas deferens* in men. These men might previously not have been diagnosed as having cystic fibrosis. With the advent of genetic testing, a specific genetic cause of their infertility or pancreatitis can be established. Genetic testing can thus have a profound impact on the diagnosis of health problems.

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6 New research, however, suggests that some people carry the gene without developing the disease, see the studies cited by J. Beckwith & J.S. Alper, "Reconsidering Genetic Anti-Discrimination Legislation" (1998) 26 J.L., Med. & Ethics 205 at 208, note 25.

7 See National Institute of Mental Health, *Genetics and Mental Health: Report of the National Institute of Mental Health's Genetics Workgroup* (NIH Publication No. 98-4268) (National Institutes of Health, 1998) at 6.

8 J. Zielenski, "Genotype and Phenotype in Cystic Fibrosis." (2000) 67:2 Respiration 117-33.

> *It should be said that the term genetic diagnostic test is also used more broadly to define all genetic tests aiming at the identification of the 'genetic status' of specific individuals, as contrasted with genetic screening.* [9]

Genetic screening

This refers to those tests that are conducted on populations with the aim of determining which individuals are sufficiently at risk of having a specific disorder so that further, more specific testing should be undertaken. These tests therefore must be sufficiently specific to allow some form of definite diagnosis, including genetic diagnosis, that may warrant therapeutic intervention. This would include, for example, pre-symptomatic testing such as BRCA1&2 testing, which may be the basis for a decision to undergo a preventive mastectomy.

Carrier testing

This is conducted to find out whether a person is carrying one copy of a recessive genetic disorder. Carrier testing can assist couples in making reproductive decisions, since it allows them to determine the risk that their offspring will inherit two copies of a mutant gene and thus be at risk for developing the condition.

Susceptibility testing

This can refer to testing that leads to the identification of a genetic mutation that makes people more susceptible to developing a disease when exposed to certain environmental hazards. For example, certain tests can identify those people who carry the gene for ataxia telangiectasia. They are more likely to develop cancer when exposed to high levels of radiation. This form of susceptibility testing will likely become a focus of debate in the context of employment.[10] *Susceptibility testing* has also been used to refer to detecting genetic mutations that indicate an increased likelihood of developing a condition such as Alzheimer's. The difference with *pre-symptomatic genetic testing* would consist here of the lower level of predictability. Finally, *susceptibility testing* can also refer to tests that can identify whether a person is more likely either to respond well to particular drug treatments, or to suffer from more severe side-effects. This form of susceptibility testing is related to a

[9] See e.g. Glass, *et al.*, *supra* note 5 and references in endnotes.

[10] For a more detailed discussion, see T. Lemmens, "'What about your genes?' Ethical, Legal and Policy Dimensions of Genetics in the Workplace." Politics and the Life Sciences (1997) 16(1): 57-75.

new area of research, pharmaco-genomics, which offers prospects of more individually tailored drug treatments.[11]

2.5 Identification Purposes: Forensic DNA and Military DNA banks

DNA is now widely used to identify tissue samples such as hair, skin particles, blood and so on, left at the scene of a crime or found attached to clothing, vehicles or other instruments used by potential suspects. The technique used to match the DNA of these samples with the DNA of identified suspects or victims is different from DNA sequencing undertaken for health care purposes. It is also unlikely that tissue or samples from a crime scene could be used to identify genetic traits or conditions. The way these samples are collected, and the often minimal amount of usable DNA that is discovered in this way makes these samples unfit for uses other than identification. Issues raised by the use of forensic samples are therefore often very specifically connected to criminal law and evidence. Nevertheless, while crime scene samples may not be fit for uses other than mere identification, law enforcement agencies have started to establish DNA banks of convicted offenders, missing persons and unsolved cases as well as population frequency databases for comparison.[12] Samples in these databases, collected in more clinically reliable circumstances, could be used for further testing. For the same purposes of potential identification, the United States military is now one of the largest collectors of DNA samples.

2.6 Existing Samples that were Collected for Other Purposes

Although genetic testing is often conducted on samples that are collected for the purpose of a specified test, genetic information can also be extracted from sources that were not provided with that aim. A couple of different scenarios are worth pointing out:

Guthrie blood spots

Across Canada, Guthrie spots have been collected from generations of new-borns. These spots of dried blood, obtained through a little foot-prick at birth, are an excellent source of DNA. Most people are unaware that in many provinces, the cards containing these spots have been kept indefinitely and that this means that others do have a potential DNA profile of them.

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[11] See R.P. Erickson, “From ‘Magic Bullet’ to ‘Specially Engineered Shotgun Loads’: The New Genetics and the Need for Individualized Pharmacotherapy: (1998) 20 Bioessays 683.

[12] See R.S. Murch & B. Budowie, "Are Developments in Forensic Applications of DNA Technology Consistent with Privacy Protections?" in M.A. Rothstein, ed., *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (New Haven: Yale University Press, 1997) 212 at 222.

Private DNA banks

Many research centres and increasingly also private laboratories and pharmaceutical companies are setting up DNA banks. The samples in these banks are sometimes originally collected as anonymous samples, or are anonymized after collection. However, this does not mean that it would be impossible to connect these samples to their originators or family members, as we will discuss later. Other samples remain identified and are connected to clinical files for further research purposes.

Family DNA storage

Several laboratories, hospitals and research centres offer, for payment, storage services for DNA of deceased family members. DNA stored this way can be helpful for family members who may later be interested in having an assessment of specific familial risks or may want to participate in genetic research.

Insurance companies

As mentioned earlier, insurance companies have traditionally been involved in gathering information on family histories of disease. This information is kept on file and some of the information is shared with the Medical Information Bureau, a non-profit association to which more than 700 American and Canadian insurance companies subscribe. When people sign a waiver of confidentiality on an insurance application, it generally gives insurance companies the explicit right to share information with the MIB. The MIB does not store complete medical records, nor does it keep very detailed medical information on individuals. It does, however, register applicants with personal information and with a three-digit code that identifies medical factors which could affect insurability. Some state that the MIB records whether insurance has been denied, while others refute this claim.[13] While detailed genetic information will not be kept by the MIB, clusters of diseases will be represented by general codes. For example, sickle cell, thalassemia and iron deficiency, will all fall under the code which represents 'anemia'. Huntington's disease will be classified as "a disorder of the nervous system." Insurance companies have also been involved in conducting HIV/AIDS testing. It does not seem impossible that they could develop an interest in obtaining blood samples and in keeping blood samples on file for purposes of risk assessment. In a way, a small blood sample would be a very concentrated source of health information that could be consulted when claims for payment are made.

[13] For more information on the MIB, see T. Lemmens & P. Bahamin, "Genetics in Life, Disability and Additional Health Insurance in Canada: A Comparative Legal and Ethical Analysis" in B.M. Knoppers, ed., *Socio-Ethical Issues in Human Genetics* (Cowansville: Yvon Blais, 1998) 115 at 168 and references there.

Immigration

Although there are no reports of immigration services using or keeping the results of specific genetic tests on file, some form of genetic information may be part of the health files of people who applied for landed immigrant status. Considering the highly predictive nature of some genetic tests and the potential implications for future health care costs, it does not seem implausible that some would defend the use of genetic testing in the context of immigration and the storage of DNA. Similar proposals have been made with respect to HIV/AIDS testing but are hopefully shelved after vocal criticism by various groups. Genetic testing has been used in immigration cases to determine parental links.[14]

Employment

In the context of employment, there are no reports of systematic compilation or use of genetic information, but it seems plausible that some genetic information may already be part of health files of employees. In the future, the further development of employment related genetic tests may push occupational health agencies and employers to store genetic information on individual employees.

2.7 Genetic Information Distinguished According to the Way it is Stored

Genetic information can also be distinguished on the basis of the way it is kept and expressed. As already suggested, genetic information can be contained in a blood sample, which then has to be further analyzed in order to release any of its secrets. Forensic DNA can be retraceable from objects, tissue or hair samples collected at crime scenes and stored by law enforcement agencies. Genetic information such as gene sequences can be available on paper or on computer files. It can be kept as a printout of a strip of DNA. Or it can be written out in the format of the sequences containing the four letters representing the chemical compounds that make up DNA. A family linkage study can be expressed in the form of a family diagram. Finally, results of genetic tests can be written down in medical files.

[14] See for example, L.T. Kirby, *DNA Fingerprinting: An Introduction* (New York: Stockton Press, 1990) at 229.

2.8 Which Definition?

We cannot expand here on the relevance of these distinctions to the various debates on ethical, legal and social issues raised by genetic testing. But as has been argued elsewhere in more detail, regulation that defines genetic information narrowly – as information resulting from DNA analysis – may lack coherence and become unenforceable as a result of the problem of defining what falls under the regulation.[15] Privacy legislation, legislation aiming at curbing genetic discrimination, or other regulatory interventions in the context of genetic testing will have to be carefully drafted so that a narrow scope does not undermine the efficacy of such efforts. For example, if regulation of genetic testing is deemed to be desirable, it would seem odd to regulate only DNA testing, and leave protein testing unregulated. Similarly, if limits on the use of genetic testing for insurance purposes are to be introduced, these should not single out pre-symptomatic screening while ignoring susceptibility testing and the specific issues raised by the advent of pharmaco-genomics.

Several earlier initiatives in other countries can be cited in this context. Belgium introduced a new insurance statute in 1992 in which the use of "genetic data" for insurance purposes is prohibited.[16] The statute specifies that a medical examination in the context of insurance can only be based on medical history and not "on techniques of genetic investigation that aim at determining a person's future health situation."[17] An Austrian federal law of 1994 also contains a sweeping prohibition on the use of the results of genetic testing by insurers as well as employers.[18] There is nothing in these statutes that clarifies how and why genetic information other than genetic test results, which is often revealed when establishing a medical history, will be excluded from further consideration by insurance companies. The Council of Europe prohibits very generally "discrimination against a person on grounds of his or her genetic heritage" and as has been argued elsewhere, this sweeping prohibition might also be untenably wide.[19] A Norwegian Act on biotechnology is somewhat more precise. While it also contains a general prohibition on the use of genetic information

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15 T. Lemmens, “Selective Justice, Genetic Discrimination and Insurance: Should We Single Out Genes in Our Laws?” McGill Law Journal (2000) 45: 347, in particular at 367-9 [hereinafter "Selective Justice"]; M.S. Yesley, “Protecting Genetic Difference” (1999) 13 Berkeley Tech. L.J. 653 at 659-62; and J. Beckwith & J.S. Alper, “Reconsidering Genetic Anti-Discrimination Legislation” (1998) 26 J.L., Med. & Ethics 205 at 207-208.

16 Wet 25 juni 1992 op de landsverzekeringsovereenkomst, B.S. 20 August 1992, in particular art. 5.

17 *Id.* art. 95 [translation: TL].

18 Federal Law of 1994 (BGB 1. No. 510/1994) *regulating work with genetically modified organisms, the release and marketing of genetically modified organisms, and the use of genetic testing and gene therapy in humans* (the Gene Technology Law) *and amending the Product Liability Law* (1995) 46 Int'l Dig. Health Legis. 42, art. 67.

19 Council of Europe, *Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine*, E.T.S. No. 164 (1997). For an analysis of its provisions and further references, see *Selective Justice*, *supra* note 15 at 357-360.

resulting from genetic tests, it makes an exception for diagnostic information resulting from genetic tests.[20] But as our discussion of the definitions above indicate, even this precision can hardly be called satisfactory. 'Diagnostic information' cannot be separated easily from other types of genetic information. And it is also not clear whether such a distinction is always appropriate in the context of insurance. George Annas, Leonard Glantz, and Patricia Roche try to give a more precise definition of genetic data in their widely cited *Draft Genetic Privacy Act*, but are thereby limiting the scope of their draft act to a degree that would make any legislation based on this model ineffective and unfair.[21] The *Draft Privacy Act* defines genetic information narrowly as information derived from DNA analysis. The authors defend their exclusion of protein tests and family history-based genetic information by referring to the need for a tight focus of their draft act.[22] Interestingly, in an article discussing the drafting of this act, they argue that including other genetic information in their proposal would have undermined the distinction between genetic information and other health information.[23] This raises the next question that we want to address. Leaving aside for a moment the difficulty of defining what exactly constitutes genetic information, and accepting the fact that the line between 'ordinary' health information and genetic information is often blurred, are there still some distinct issues raised in the context of the increasing use of genetic information?

[20] *Law No. 56 of 5 August 1994 on the medical use of biotechnology* (1995) Int'l Dig. Health Legis. 51, s. 6-7.

[21] For a more detailed discussion, see "Selective Justice", *supra* note 15 at 367-368.

[22] G.J. Annas, L.H. Glantz & P.A. Roche, *The Genetic Privacy Act and Commentary* (Boston: Boston University School of Public Health, 1995).

[23] G.J. Annas, L.H. Glantz & P.A. Roche, "Drafting the Genetic Privacy Act: Science, Policy and Practical Considerations" (1995) 23 J.L., Med. & Ethics 360.

3. CHARACTERISTICS OF GENETIC INFORMATION: THE CLAIM OF GENETIC EXCEPTIONALISM

Our discussion of the different forms of genetic information already makes clear that we are not necessarily dealing here with something totally new. Genetics has been around for some time in one form or another. But that does not necessarily address the arguments invoked to defend the exclusive nature of genetic information. Indeed, it could very well be that the exclusivity of genetic information was less of an issue in the past because it was less widely used. In other words, it still could be true that genetics is truly distinct and merits a distinct regulatory approach. We therefore have to address the arguments invoked to support 'genetic exceptionalism.' Lawrence O. Gostin and James G. Hodge define this as "the societal practice of treating genetic data as different from other types of health data for the purposes of assessing privacy and security protections."[24] We will indicate first why genetic information shares most, if not all, of its characteristics with other types of health information. We will then concentrate on those aspects of genetics that have caused many to be more concerned about its use now than, say, thirty years ago. As already suggested, we will argue that the fear for abuse is connected not so much to the inherently new characteristics of genetics, but rather to the link between some of its shared characteristics and the advent of new computer technology.

In a fascinating chapter in which he also discusses the *Draft Genetic Privacy Act*, philosopher Thomas Murray analyzes four different arguments invoked to support "genetic exceptionalism": 1. Genetic prophecy 2. Concern for kin; 3. Concern about discrimination; 4. Generalizability of data to families, communities, racial and ethnic populations.[25] Discussing his analysis in an article in the *McGill Law Journal*, one of us also addressed a fifth characteristic often invoked in discussions about genetics: the argument about lack of control over one's genome.[26]

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[24] Lawrence O. Gostin & James G. Hodge, "Genetic Privacy and the Law: An End to Genetics Exceptionalism" (1999) 40 Jurimetrics 21 at 31 [hereinafter "Genetic Privacy"].

[25] T. Murray, "Genetic Secrets and Future Diaries: Is Genetic Information Different from Other Medical Information?" in Rothstein, *supra* note 12, 60-73 [hereinafter "Genetic Secrets"].

[26] See "Selective Justice" *supra* note 15 at 370. The following discussion is largely based on the analysis there. See in particular 369-380.

3.1 Genetic Prophecy

'The concern for genetic prophecy' refers to the predictive character of genetics. Most genetic tests reveal a risk factor without necessarily saying anything about actual health. They give us a snapshot of what can or, in some cases even is likely to happen in the future to our health. Genetics would differ substantially from other health information obtained in 'ordinary' medical examinations, according to this argument, because the latter only give us information about current health. Third parties such as insurers, employers, immigration authorities and perhaps even judicial authorities have, for various reasons, an innate desire to know more about the future health status, behavior or performance levels of employees, immigrants or convicts. They could be interested to use this genetic technology in a way that may harm these individuals.

However, as Murray and others indicate, information on future health can and has been obtained from various other sources: family histories of disease, various medical tests indicating high blood pressure, cholesterol levels or iron levels in blood have been widely used in medicine for decades.[27] Eating habits, life-style and often even physical appearance will give others some clues about our future health. Tests for HIV/AIDS and hepatitis have been around for some time. With the advent of new forms of treatment for HIV/AIDS, a positive test result for HIV/AIDS is no longer an imminent death sentence. People are surviving longer with the infection. They are in a situation that is to some extent similar to some people who test positive for cystic fibrosis. They may suffer greatly from their condition and may be seriously limited in their daily life as a result of the disease, but they are often capable of controlling the progress of the disease.

Moreover, some people seem to be immune to HIV-infection. Others who are infected with HIV do not develop full-blown AIDS, or only much later. Specific genetic mutations are likely the reason for the variable expression of the disease.[28] This brings us to a point that we already mentioned in the discussion of the various forms of genetic information: genetic research is increasingly showing us how variable the impact of genetic factors on disease really is. For example, studies now indicate that some people who test positive for Huntington's disease do not develop the disease.[29] With respect to cystic fibrosis, the disease has shown to be much more complex than originally conceived and involves not only one

[27] "Genetic Secrets", *supra* note 25; see also the discussion in Alper & Beckwith, *supra* note 3.

[28] See "Genetic Privacy", *supra* note 24 at 32 and references there.

[29] See Beckwith & Alper, *supra* note 15 at 208, and references in note 25.

common mutation, which accounts for about 70% of the disease, but also a staggering number of more than 850 different mutations.[30] Many differences in phenotypic expression of the disease are associated with these different mutations. Environmental and secondary genetic factors (interaction with other genes) are now considered to have an impact on cystic fibrosis. This affects the predictive power of the genetic tests for cystic fibrosis. In a way, this pre-symptomatic testing for cystic fibrosis is in these cases not fundamentally different from susceptibility testing. For other diseases, it is even clearer that environmental factors and gene-gene interactions play a major role in their development.

It is fair to say, then, that the risk factors associated with specific genetic mutations and the predictive power of the existing genetic tests vary greatly. The severity of the disorders associated with one or more known genetic mutations is very diverse. The traditional severe single-gene disorders are rare in comparison with the variety of conditions that have now been identified as being associated with specific genes. Moreover, a severe disease such as cystic fibrosis, which was previously untreatable, has become more controllable. Cystic fibrosis also highlights how genetic disorders can be expressed variably, thus making prediction of future health more uncertain. Genetic disorders, even the very determinant ones, are often characterized by "incomplete or reduced penetrance" meaning that the time of onset of the disease varies.[31] Other diseases can be avoided or their impact reduced by changes in diet or lifestyles. In other words: it seems difficult to defend the uniqueness of genetics on the basis of a commonly shared predictive character of the information provided by genetic tests. With respect to predicting future health, there is too much variability in genetic tests to support that claim.

It is also important to keep in mind how later research often corrects earlier overstatements about the predictive character of specific genetic tests. We mentioned the case of Huntington's disease and cystic fibrosis. As discussed elsewhere, BRCA1 and BRCA2 tests for breast cancer are also good examples of how later findings may tone down the predictive power of genetic tests.[32]

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[30] See J. Zielenski, "Genotype and Phenotype in Cystic Fibrosis." (2000) 67:2 Respiration 117-33; in another publication, one of us invoked the number of 550 mutations, but referred to a 1996 publication. ("Selective Justice", *supra* note 15at 379.) This simply highlights the rapid pace of discoveries in this area.

[31] For a more detailed discussion, see Lemmens & Bahamin, *supra* note 13 at 135-140.

[32] "Selective Justice," *supra* note 15 at 371 and references there.

3.2 Lack of Control Over One's Genome

The lack of control over one's own genome is sometimes invoked to differentiate genetics from some other types of health information. In discussions about the need to protect people against third party use of genetic information, some state that it would be unfair to use the presence of certain genes to the detriment of those who carry them, because genetics escapes individual control.[33] Indeed, people do not choose their genetic make-up; they are born with it. Or, as Gostin and Hodge frame it, "[p]eople feel stuck with their genes."[34] Several points should be raised about this argument to show that it cannot be invoked to differentiate genetics from other health information. First, if GATACCA-like predictions come true, individuals will increasingly be able to select perhaps not their own genes, but certainly the genes of their offspring through carrier screening and pre-natal diagnosis. The GATTACA "pre-mating ritual" of having a genetic screen of one's partner clearly is a caricature of what is going on now, but some selection of traits does take place. Various methods are used to obtain certain traits in offspring or to avoid other traits. Results of genetic tests are used to inform couples about their risk for giving birth to children with genetic disorders. People who select sperm for artificial insemination do select to some extent half of the genes of that baby through selecting the profile of the sperm donor. If control is the morally relevant criterion to assign blame, then parents may increasingly be held accountable for the genetic make-up of their children since they partly selected it.

Second, as Gostin and Hodge point out, "[g]enetic flaws, like environmental diseases, can increasingly be altered or corrected through clinical interventions."[35] Developments in gene therapy will bring genetic traits more under control, even for the living who are 'stuck with them.' Moreover, as we pointed out in the introductory part, genetic testing will often only refer to an increased susceptibility, and individuals who test positive for such susceptibilities may be able to control the onset of disease through changes in life-style and other preventive measures.

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[33] See among others M.A. Rothstein, "Genetics, Insurance and the Ethics of Genetic Counseling" (1993) 3 Molecular Genetic Medicine 159 at 169 and R.S. Brown, "The impact of Advances in Genetics on Insurance Policy" in R.S. Brown & K. Marshall, eds, *Advances in Genetic Information: A Guide for State Policy Makers* (Lexington, Ky.: Council of State Governments, 1992) at 47.

[34] "Genetic Privacy", *supra* note 24 at 34

[35] *Ibid.* (footnote omitted).

Third, the idea that genetics is different and merits special protection because it is 'beyond our control' may contribute to some extent to claims of genetic determinism. Genetic determinism refers to the belief that everything, including human behavior, is ultimately determined by people's genetic structure.[36] If one were to say that genetics has to be protected because it is not controlled, it could contribute to the perception that genes really do fully determine who we are and what kind of life we may expect. In a way, even if well intended, regulatory initiatives in this area, if justified by reference to lack of control, could contribute to an erroneous public perception of genetics.

Moreover, there is also considerable debate about the appropriateness of using the notion of 'control' to assign moral blameworthiness. What does it mean to have control over one's health? People who have control over their health and still 'choose' to become sick or disabled by unhealthy life-styles or risky behavior, according to this mode of thinking, may deserve less protection and less access to health care.[37] But what constitutes choice? How do we decide on what constitutes a morally relevant contribution to disease and disability? One would not want to argue that, for example, battered women are more responsible for their health risks than someone who has a susceptibility to cancer and does not manage to follow a risk-reducing diet.[38] As one of us argued elsewhere, "[t]o hold people accountable for "lifestyle-related" increased health risks without taking into consideration the social, cultural and environmental context is, in many cases, to further discriminate against populations already vulnerable to the negative effects of discrimination."[39] Furthermore, research in behavioral genetics suggests that genetic factors may contribute to some of these 'lifestyle choices' such as alcoholism and nicotine addiction, or even engaging in risky sports.[40] Using some form of genetic determinism as the basis for distinguishing genetically 'caused' from 'self-inflicted' diseases thus creates a stark contradiction. If genetic determinism holds true, then those self-inflicted behaviors are equally beyond our control.

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[36] For a short discussion of genetic determinism, see Bartha Maria Knoppers, *Human Dignity and Genetic Heritage: A Study Paper Prepared for the Law Reform Commission of Canada*, Protection of Life Series (Ottawa: Law Reform Commission of Canada, 1991) at 43-46.

[37] For a recent analysis, see A. Darby, "The individual, health hazardous lifestyles, disease and liability" (1999) 4 DePaul Journal of Health Care Law 787, in particular at 798-807.

[38] For the debate about battered women and the fairness of insurance underwriting for these women, see D. Hellman, "Is actuarially fair insurance pricing actually fair? A case study in insuring battered women." (1997) 32 Harvard Civil Rights-Civil Liberties Law Review 355.

[39] "Selective Justice" *supra* note at 375.

[40] See J. Beckwith & J.S. Alper, "Human Behavioral Genetics" (1996) 10(2) The Genetic Resource 5. A tendency for risk-taking behaviour could be associated with research suggesting a link between a specific genetic marker and "novelty-seeking" behaviour. See *id.*at 8 and references there.

Finally, our discussion of this issue should not be seen as an endorsement of the moral relevance of this distinction in the first place. Indeed, a just society may very well have to disregard the fact that one is partly responsible for one's illness and expand a protective gaze to embrace all those who suffer from illness and poverty, irrespective of its cause.

3.3 Family and Ethnic Community

The arguments about *family relevance* and *ethnic relevance* of genetics are more interesting and more complex to discuss. *The concern for kin*, as Murray phrases the former, refers to the way genetics links us to our families. Genetic tests necessarily reveal information about family members. Children of a parent who tests positive for Huntington's disease, a dominant genetic disorder, have a 50% chance of inheriting a copy of the mutant gene and thus of developing the disease. Clearly, a positive test result of a parent also provides information about the risk status of his or her child. Children from parents who both carry the thalassaemia gene have a 25% chance of inheriting the two mutant copies from their parents (one from each parent). If a woman diagnosed with breast cancer subsequently tests positive for a known disease causing mutation of the BRCA1 or BRCA2 gene, then her first-degree relatives (children, siblings) have a 50% likelihood of carrying the same mutation and therefore of having an increased cancer risk.[41] In particular, when it comes to potentially preventable or treatable conditions, it can be very important for people to be informed of a family member's genetic test results and thus of their own risk. Some types of genetic research (genetic linkeage studies) are impossible without the participation of family members. These factors raise questions with respect to the duty of physicians and patients to inform or disclose genetic risk information to family members, family members' right not to know this information, and family members duty to collaborate in research.[42] Other family issues are also raised in the context of genetics. DNA techniques allow for an accurate determination of parentage. This can be particularly important in paternity disputes. It has been used in courts, for example in child support and immigration cases.[43] The wide-spread

[41] Caryn Lerman, Beth N. Peshkin, Chanita Hughes, Claudine Isaacs, "Family Disclosure in Genetic Testing for Cancer Susceptibility: Determinants and Consequences" (1998) 1 Journal of Health Care Law and Policy 353 at 355 [hereinafter "Family Disclosure"].

[42] For a discussion of these issues, see *e.g. Whose Hands on Our Genes*, *supra* note 5, in particular at 15; E.W. Clayton, "What Should the Law Say About Disclosure of Genetic Information to Relatives?" (1998) 1 Journal of Health Care Law & Policy 373; C. Lerman *et al*. "Family Disclosure in Genetic Testing for Cancer Susceptibility: Determinants and Consequences" (1998) Journal of Health Care Law & Policy 353; M.M. Burgess, C.M. Laberge & B.M. Knoppers, "Bioethics for clinicians: 14. Ethics and genetics in medicine (1998) 158 CMAJ 1309; W.F. Flanagan, "Genetic Data and Medical Confidentiality" (1995) 3 Health L.J. 269.

[43] *Whose Hands on Our Genes*, *supra* note 5 at 13; an immigration case is discussed in Kirby, *supra* note 14 at 229.

use of non-paternity testing clearly undermines the value of the legal 'presumption of paternity' as social stabiliser within marital relations – provided it still has this value. It changes the way paternity issues can be dealt with. Family secrets may now be brought into the broad daylight without the consent of the mother involved.[44] Genetic testing may also open the door for many orphans and for children created by artificial insemination to retrace their biological parents, even when clear adoption or donor records are missing. Genetic testing can also open the books on generation-old myths of parentage. For example, DNA testing is being used to assess heritage claims related to various historical figures such as former U.S. president Jefferson.[45] This raises interesting questions about whether and to what extent we ought to respect people's desire to keep biological secrets after death.

Non-paternity will more often be discovered by accident during routine clinical testing or genetic research. When searching for specific genes, family linkage studies are often conducted to determine how specific markers for genetic diseases are inherited across families. Non-paternity may also be discovered during routine genetic testing when researchers or clinicians simultaneously test samples of different family members. In these cases, researchers or clinicians may face a dilemma between their duty of confidentiality towards the parent involved, and their duty of disclosure towards the children. In practice, it appears that clinicians and researchers do not always warn family members of the risk of discovering non-paternity and do not discuss with them the option of non-disclosure. When non-paternity is discovered, they may withhold that information. This creates the odd situation whereby physicians or researchers may not be willing to tell individuals the real results of their tests (e.g. the real reason why they are not at risk for developing a disease running in the family).

While there are certainly important familial issues surrounding genetics, we would argue, as Murray does, that family relevance of genetic information *per se* does not make genetic information unique. As we pointed out, genetic testing has created some interesting issues with respect to how families can be affected by this information, and how they have to deal with it, but family relevance itself is not a distinguishing feature. The difference, so we shall argue, lies not so much in the familial nature of the information, but in the level of detail and in the way that the familial aspects of genetics may play out in some areas of the law.

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[44] The Human Genetics Commission report points out that new genetic tests no longer require the participation of the mother. These 'motherless' tests can be performed, for example, with DNA provided by a doubting father and his child. *Whose Hands on Our Genes*, *supra* note 5 at 14.

[45] Sean Wilentz, "Hemings Hawing" [editorial] New Republic (Nov. 30, 1998) 16.

Genetic information is not unique in disclosing information about other people. Other forms of health information also do so. For example, our medical files often include information about our family members as physicians routinely ask questions about family background. Some of this may be genetic information in that it discloses familial pre-dispositions to particular conditions. But some of this may be non-genetic information regarding issues such as past and present lifestyle, mental and sexual health.

Indeed, there are many other types of information that affect families. The physical and social environments of families tell us something about all their members. When one person develops cancer because of exposure to toxins in the family home, others know that they are at increased risk. Tuberculosis in one family member tells us something about the risk of people living with them. Alcoholism of one family member may be an indicator of risks for other family members' health and wellbeing. Gambling problems affect the family's financial security. And if the "breadwinner" has a serious life-threatening illness, Murray argues, surely other family members have an interest in knowing about it. One comparison that is often made when it comes to discussing the duty to disclose genetic risks, is the comparison with disclosure of sexually transmitted diseases to partners.

Even the case of not divulging the 'true' results of genetic testing is not necessarily new. Family secrets have been divulged in the past to physicians, psychiatrists, and other professional confidants. In some cases, this may have been relevant for family members' health care choices but physicians have traditionally respected their duty of confidentiality.

Another characteristic of genetics is that it may affect or tell us something particular about *ethnic, racial or local groups*. Genetic information is to some extent shared between larger communities. The most traditional examples of community relevance of genetic information relate to the higher presence of specific diseases within some ethnic groups, or in specific local communities. Tay-Sachs, for example, is common among Ashkenazi Jews and some French Canadians.[46] Ashkenazi women who have a family history of breast cancer are at a higher risk of developing cancer than women of other ethnic groups.[47] Sickle-cell trait, which is also associated with a higher resistance against malaria, has a very high incidence among Africans and African-Americans. From 8% to 10% of African-Americans are carriers of the

[46] *Genetics in Canadian Health Care*, *supra* note 5 at 42.

[47] See S.V. Hodgson *et al.*, "Risk factors for detecting germline BRCA1 and BRCA2 founder mutations in Ashkenazi Jewish women with breast or ovarian cancer" (1999) 36(5) Journal of Medical Genetics 369; see also Rothenberg, *supra* note

sickle-cell trait, and 1 in 400 to 600 has sickle-cell anaemia.[48] Cystic Fibrosis is more common among Caucasians.[49] When research leads to the development of genetic tests for conditions that are affecting a particular ethnic community, members of this community may be stigmatized. The development of better knowledge about this particular genetic disease and the publicity that often comes with new discoveries may attract attention and may lead to increased discrimination. Often, these communities are already stigmatized and affected by discrimination. Genetic testing could then become yet another, more sophisticated tool of discrimination. In the United States, sickle-cell screening programs and erroneous scientific interpretations about the risk of being a carrier of the trait, lead to discrimination against African-Americans.[50] Behavioral genetics research linking intelligence, criminality, attention deficit hyperactivity disorders, and other behavioral traits to specific genes that may be more prevalent in some ethnic communities than in others creates even greater risks for stigmatization and discrimination. It is also interesting to note that some communities have participated very intensively with genetic research. This genetic research has, in turn, often been of benefit to the general population. It seems unfair that those who contributed in this way would also become particularly vulnerable as a result of their participation.

New DNA technologies have also contributed to various types of research with ethnic or population relevance. DNA research may impact on the cultural identity of these communities. For example, DNA research on the South-African tribe of the Lemba confirmed their claims based on oral tradition and respect for Jewish religious rules that they are direct descendents of Jews led out of Judea by their religious leader Buba. Genetic researchers found that many Lemba men carry a set of DNA sequences that is distinctive of the Cohanim, the Jewish priests believed to be the descendants of Aaron. This DNA sequence is particularly common among those Lemba men who, according to their tradition, are also members of a priest-like clan.[51] DNA research is increasingly used to determine the migratory history of people. This research has significant archeological and historical value. Some stress it could also be a social tool, since it shows how much different ethnic groups have in common. At the same time, it could also raise sensitive issues about our ancestors and culture. What if DNA research is used to rebut land claims of aboriginal groups based on

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[48] According to the Science Council of Canada, one in every 625 black newborns has sickle cell-anaemia. (*Genetics in Canadian Health Care*, *supra* note 5 at 20.)

[49] *Id.* at 18.

[50] See D.J. Kevles, *In the Name of Eugenics: Genetics and the Uses of Human Heredity* (Berkeley: University of California Press, 1985) 255-256 and 278; and L.B. Andrews *et al*. (eds), *Assessing Genetic Risks: Implications for Health and Social Policy* (Washington: National Academy Press, 1994) at 40-42 and 258.

[51] N. Wade, "DNA Back South Africa Tribe's Tradition of Early Descent from the Jews" New York Times (May 9, 1999)

their right of first arrival? Or if DNA testing is used to determine whether someone truly is a member of an aboriginal group? In the United States, compensation offered by the United States' government created tension between different members of the Seminole Nation. Descendants of escaped slaves who for generations considered themselves members of this Nation and seem to have been accepted as such were excluded from those who would receive compensation on the basis of blood lineage.[52] Similar debates take place elsewhere, including in Canada, and it does not seem impossible that some would call for the use of genetic in this type of debate.

The impact of genetic tests on communities raises important questions with respect to obtaining informed consent, respecting community values and practices, the need to involve communities in the design, conduct and analysis of research, and so on. [53] Genetics has certainly created particular dilemmas for communities and for researchers conducting research with these communities. However, concerns about the impact of research on communities are again not unique to genetics. Statistics indicate differences in the incidence of cancers among local communities,[54] the lower incidence of high cholesterol levels among certain ethnic groups, the fact that HIV/AIDS is more prevalent among gays, intravenous drug users and specific ethnic communities, and so on. Postal codes are used as indicators of a higher chance of being infected by HIV/AIDS, or of bad housing and living conditions which may affect one's life expectancy. Poverty rates affect particularly certain ethnic communities, such as aboriginal peoples in Canada and African-American communities in the big American cities. Poverty is the most direct indicator of low life expectancy and ill health. Research on, for example, alcoholism in communities and life style, can affect and stigmatize the community that is the target of research.

[52] W. Glaberson, "Who Is a Seminole, and Who Gets to Decide?" New York Times (January 29, 2001).

[53] For an overview of ethical issues raised by research involving communities and for an analysis of existing regulations, see C. Weijer, G. Goldsand, E.J. Emanuel "Protecting communities in research: current guidelines and limits of extrapolation." (1999) Nature Genetics 23: 275-280; C. Weijer & E.J. Emanuel, "Protecting communities in biomedical research (2000) Science 289: 1142-1144; see also a short discussion in Glass *et al.*, *supra* note 5.

[54] Lappé, M.A. "Justice and the Limitations of Genetic Knowledge" in T.F. Murphy & M.A. Lappé, eds., *Justice and the Human Genome Project* (Berkeley: University of California Press, 1994) 153 at 156.

4. HOW GENETICS HIGHLIGHTS EXISTING PROBLEMS

So far, we have argued that genetic information shares various characteristics with other types of health information and that genetics does not raise inherently new ethical and legal questions. However, that does not mean that there is no need for increased scrutiny of existing regimes or the development of new regulatory responses. Three reasons justify careful analysis, public debate and regulatory intervention where needed. First, even though genetics does share many of its characteristics with other health information, that does not mean that we have found satisfactory ethical, legal and regulatory answers to address issues raised by these characteristics. More systematic use of genetics deserves special attention because it could exacerbate existing problems. Murray claims that the increasing number of genetic tests "broadens the pool of possible factors that might be used to discriminate against an individual, and it likewise increases the number of individuals who might become the subjects of discrimination."[55] It is worth pointing out that significant ethical and social problems could also be created by the fact that these 'exacerbated problems' increasingly play out in a much more commercial health care context. Second, genetics brings many of these issues to a different level. As we argued in the context of the familial impact of genetics, genetics gives a new spin to some of our concerns, which may require a different regulatory approach. In our analysis, we have given examples of the way genetics brings these issues up to a different level: genetics gives us more detailed risk information than most other tests; genetics increases dilemmas surrounding the duty to inform third parties such as family members of particular health risks; and genetics does impact in particular ways on communities. Third, a combination of the following factors, which are not necessarily "unique characteristics" of genetics, makes it necessary to look at how adequate current regulatory approaches are: 1) the volume of information that can be extracted from one sample which can be kept indefinitely; 2) the speed of testing; and 3) the link between genetics and computer technology. This third issue is clearly related to the first two issues and is contributing to the ease by which genetic information can be obtained.

For the remainder of our paper, we want to analyse some of these elements further. First, we will analyse in more detail how genetics may shed new light on an existing issue. We will use as an example the issue of duty to inform others of health risks. We will then discuss how the nature of a genetic sample, in particular the volume of information contained in it and its

[55] "Genetic Secrets", *supra* note 25 at 66.

potential longevity raises particular issues. We will conclude by discussing how computer technology impacts on this debate.

4.1 Family and Disclosure of Risk Information

We have already indicated that the fact that an individual's genetic information also reveals information about biological family members, raising concerns regarding the disclosure of such information to relatives, does not render genetic information unique. Other information may also be of concern to an individual's relatives. However, characteristics of genetic information and the way in which it is relevant to third parties calls into question traditional justifications for disclosure or non-disclosure of health information to third parties and suggests that there may be a need for unique guidelines for the disclosure of genetic information.

There is considerable controversy over whether a physician has a privilege (or, more strongly, a duty) to breach duties of confidentiality in order to warn a patient's relatives as to their risk of a genetic condition.[56] The breach of confidentiality involved in disclosing a patient's health information against his or her wishes is generally only justified where there is a threat of serious and imminent harm to others or there is a serious public health risk involved.[57] An example that would fall into this traditional rationale would be infectious disease. When an individual has an infectious disease this is of concern to that individual's family (and those who come into close contact with him or her) because it alerts them to a potential risk to them arising from their physical and social environment. Genetic risks are to some extent different. For example, as we pointed out, if a woman diagnosed with breast cancer subsequently tests positive for a known disease causing mutation of the BRCA1 or BRCA2 gene, then her first-degree relatives (children, siblings) have a 50% likelihood of carrying the same mutation. They therefore have a significantly increased cancer risk.[58] But

[56] See e.g., *Pate* v. *Threlkel*, 661 So.2d 278 (Fla. 1995) (physicians have a duty to warn of a genetically transferable disease, but they only have to inform their patients, not their family members) and *Safer* v. *Estate of Pack*, 291 N.J. Super. 619 A.2d 1188 (1996) (physicians can have a duty to warn relatives about genetic conditions). For a case comment on the latter, see A. Liang, "The Argument Against A Physician's Duty to Warn for Genetic Diseases: The Conflicts Created by *Safer v. Estate of Pack*" (1998)1 Journal of Health Care Law & Policy 437.

[57] The cases generally involve infectious diseases, where disclosure to public authorities is often mandated by legislation such as Ontario's *Health Protection and Promotion Act*, R.S.O. 1990, c.H.7, or cases pertaining to mental health professionals who were held to have knowledge that their patient posed a serious risk of violence to another, such as in the famous case of *Tarasoff* v. *Regents of University of California* 551 P.2d 334 (Cal. 1976). See also *Jones* v. *Smith* [1999] 1 S.C.R. 455 (public safety can in some circumstances outweigh solicitor-client privilege attaching to a psychiatric affidavit held by the defence).

[58] C. Lerman, B.N. Peshkin, C. Hughes, C. Isaacs, "Family Disclosure in Genetic Testing for Cancer Susceptibility: Determinants and Consequences" (1998) 1 Journal of Health Care Law and Policy 353 at 355 [hereinafter "Family Disclosure"].

she has not *created* the risk for the relative. It is a *preexisting* risk that is identified because of her genetic information.[59] The risk is not so much related to something external to the other person. It is a risk associated with genes that are part of that person. This distinguishes genetic information from the type of harm involved in the recognized exceptions to the duty of confidentiality.

Furthermore, the harm disclosed by a pre-existing genetic condition is not necessarily preventable. If it is not preventable then disclosure likely will not help the third party and may even harm the third party by causing distress. To the extent that the manifestation of the disease associated with a particular genetic condition is preventable, because of complicating biological and environmental factors, then it looks a lot less like the serious and imminent harm at issue in the traditional justification for a breach of confidentiality (even though the information is in that case so much more relevant for the family member).

There are further important questions that need to be answered regarding what counts as a serious or imminent harm in this context. The first is the degree of likelihood of having a genetic condition. That is, is a 50% likelihood of carrying a mutation of the BRCA1 gene sufficiently serious? The second element is the degree of likeliness that the genetic predisposition will lead to the actual physical manifestation of the disease. In other words, if having a particular mutation is associated with a 63% increase in the likelihood of a particular condition then is this sufficiently serious?[60] Does it matter that this represents the known lifetime risk, rather than some imminent health risk?

The Science Council of Canada has adopted the same guidelines for physician disclosure to third parties as the President's Commission for the Study of the Ethical Problems in Medicine and Biomedical and Behavioral Research:

1. reasonable efforts to elicit voluntary consent to disclosure have failed;
2. there is a high probability both that harm will occur if the information is withheld and that the disclosed information will actually be used to avert harm;
3. the harm that identifiable individuals would suffer would be serious;

[59] L.B. Andrews, "The Genetic Information Superhighway: Rules of the Road for Contacting Relatives and Recontacting Former Patients" in B.M. Knoppers *et al*, eds., *Human DNA: Law and Policy* (1996) 133 at 138.

[60] The lifetime risk of breast and ovarian cancer associated with BRCA1 mutations are 85% and 63%. See "Family Disclosure", *supra* note 58 at footnote 1.

4. appropriate precautions are taken to ensure that only genetic information needed for diagnosis and/or treatment of the disease in question is disclosed.[61]

While these guidelines help to clarify some of the issues outlined above, questions remain.

One question is whether harm is the appropriate way to think about disclosure of genetic information to third parties. In general, an individual has a right to know his or her own genetic constitution, based on the value of individual autonomy.[62] This rationale can also be used to justify the right not to know one's own genetic constitution.[63] But this rationale would also provide a reason for biological relatives to gain access to the genetic information of a family member: it is also information about their own genetic constitutions. Therefore if they seek this information, in situations where they can only learn about their own genetic information through gaining access to information about relatives (e.g. linkage studies), then there is a reason for disclosing this information that is based on autonomy rather than harm. An autonomy rationale would provide more reasons to allow access to another's genetic information than a harm rationale. It therefore at least raises the following questions:

- If a test is performed and the individual does not want to be told of the results, can family members nonetheless access this information?
- Should a parent be allowed to seek genetic testing of a child in order to find out genetic information about herself or the child's siblings? Does this violate the child's right not to know?[64]
- Should relatives be allowed access to an individual's genetic information after that individual has died?[65]
- Should a relative be granted access to the genetic information about another only when his or her own health is at stake, or also in situations of reproductive decision-making?

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[61] *Genetics in Canadian Health Care*, *supra* note 5 at 72-73; President's Committee for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research, *Screening and Counseling for Genetic Conditions: The Ethical, Social and Legal Implications of Genetic Screening, Counseling, and Education Programs* (Washington D.C.: Government Printing Office, 1983) at 44.

[62] R. Chadwick, "The Philosophy of the Right to Know and the Right not to Know" in R. Chadwick, M. Levitt and D. Shickle, eds., *The Right to Know and the Right not to Know* (Aldershot: Avebury, 1997) 13 at 14-16.

[63] Some commentators argue that there may be situations where we would want to hold that someone is obligated to pursue genetic knowledge. See *e.g.*, R. Rhodes, "Genetic Links, Family Ties, and Social Bonds: Rights and Responsibilities in the Face of Genetic Knowledge" (1998) 23 J. Medicine & Philosophy 10.

[64] K.C. Glass, "Access to Genetic Information" in B.M. Knoppers *et al*, *supra* note 59,157 at 158.

[65] Quebec allows access to a deceased person's medical history to ascertain the existence of a hereditary or genetic disease, even if while alive that person objected to such access by others: *Act Respecting Health Services and Social Services*, S.Q. (1991) c. 42, art. 23.

- If a linkage study discloses information regarding paternity, when should this be disclosed?

Of course, there are competing autonomy interests at stake here, which are difficult to balance. It may be that the claims of relatives are not strong enough to outweigh health care professionals' duties of confidentiality. At a minimum, what these questions highlight is the need for genetic counseling services to be widely available to aid families as they deal with these issues.

4.2 Information, Longevity and Identification

An analogy can be made between genetic information and information technology. For more than two decades, concerns regarding the practices made possible by burgeoning information technology have spurred the adoption of data protection regimes. These concerns include:

> *the ubiquitous use of computers for the processing of personal data, vastly expanded possibilities of storing, comparing, linking, selecting and accessing personal data, and the combination of computers and telecommunications technology which may place personal data simultaneously at the disposal of thousands of users at geographically dispersed locations and enables the pooling of data and the creation of complex national and international data networks.*[66]

One commentator has distilled these concerns into two features of information technology: its ability to shift information from the context in which it was gathered to a different context, and its ability to aggregate data.[67]

If we deal with genetic information in the form of DNA samples, or substantial sequencing information, then similar issues are raised as in the example of information technology. The amount of information about an individual that can be gleaned from a DNA sample is potentially staggering. If the sample is stored, then it has the potential to reveal information

[66] OECD, *Guidelines on the Protection of Privacy and Transborder Flows of Personal Data*, *Explanatory Memorandum,* Annex to Recommendation of the Council, September 23, 1980, at para.3. These guidelines have been adopted by the Canadian Standards Association in their *Model Code for the Protection of Personal Information* (1996), available at http://www.efc.ca/pages/doc/csa-privacy-code.jun96.html (on file with author) and formed the basis of Canada's *The Personal Information Protection and Electronic Documents Act*, S.C. 2000, c.5 [hereinafter *Bill C-6*].

[67] H. Nissenbaum, "Protecting Privacy in an Information Age: The Problem of Privacy in Public" (1998) 17 Law & Phil. 559.

about the individual far into the future, beyond the purposes for which the individual first provided the sample, and even long after the individual's death, limited only by available genetic knowledge and technique. Furthermore, DNA can uniquely identify an individual, which can provide the basis for linking together information from disparate sources. A clear example of this is in the forensic context, where biological material gathered from a crime site can be compared to existing "DNA fingerprints" of suspects.

As the analogy with information technology indicates, the fact that genetic information allows for the aggregation of information and its shift between contexts is shared by information technology. And, as other commentators have pointed out, the science of biometrics allows for other ways to uniquely identify an individual than by using DNA fingerprinting.[68] However, the particular contexts in which these concerns arise with respect to genetic information may be unique and warrant a specific focus, and response, when regulating health information.

For example, it is quite common, when developing data protection regimes, to exempt non-personally identifying data from protection.[69] Similarly, existing human tissue samples used in research are often exempt from consent requirements when used in non-identifiable form.[70] There are serious questions, however, regarding the extent to which genetic information can be collected or used in a non-identifiable form. Genetic information, unlike other health information, is inherently linked to a particular individual.[71] This fact, in combination with computer technology, makes the linkage of genetic information to an identifiable individual always a possibility. As the Tri-Council Policy Statement on Ethical Conduct for Research Involving Humans states, "[g]enetic testing has greatly narrowed the concept of anonymous tissue... [T]he concept of traceable tissue is now wider, since it is now possible to identify biological relatives by using genetic markers."[72]

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68 "Genetic Privacy", *supra* note 24 at 34-5.

69 See *e.g.*, *Bill C-6*, *supra* note 66 at s. 4 (Act applies to the collection, use and disclosure of "personal information" defined in s. 2(1) as "information about an identifiable individual").-

70 See *e.g.*, the (no longer in place) Medical Research Council of Canada, *Guidelines on Research Involving Human Subjects 1987* (Ottawa: Minister of Supplies and Services Canada, 1987) at 26: "consent is generally unnecessary for research undertaken, for example, upon surplus blood, urine, tissue, and similar samples obtained for diagnostic or treatment purposes if the patient is not identifiable, and the requirements of the research do not influence the procedures used for obtaining samples." For an American example, see 45 C.F.R. § 46.101(b)(4) (1998) (Department of Health and Human Services Policy for Protection of Human Research Subjects.

71 Lawrence O. Gostin "Genetic Privacy" (1995) 23(4) Journal of Law, Medicine & Ethics 320 at 322.

72 Social Sciences and Humanities Research Council, Natural Sciences and Engineering Research Council, Medical Research Council, *Tri-Council Policy Statement on Ethical Conduct for Research Involving Humans* (Ottawa: Public Works and Government Services Canada, 1998) at 10.2 [available on-line at <http://www.nserc.ca/programs/ethics/english/index.htm>]

Interestingly, the new Canadian Tri-Council Policy Statement does impose a more stringent consent requirement for collection of tissue and for the use of previously collected tissue, but it leaves the door open for the use of previously stored anonymous tissue without consent.[73] It is worth noting also that the Tri-Council Policy Statement requires researchers to specify in a research protocol future use of the genetic material. In other words: DNA banking of material should be justified at the time of collection. Moreover, the Policy Statement also suggests that there should be a time limit on the storing of samples, which should be congruent with the purpose of storage.[74]

Very often, samples collected for research purposes are not made anonymous. They can be directly identifiable through the use of an identifying tag or through patient or research subject numbers. Or they can be traced back through a specific identifying code, which may be kept at a different location and/or by a different person than the person who conducts research on the sample. Linkage between the samples and the clinical file can be of crucial importance either for clinical or for further research purposes. Researchers are worried that rules that are too stringent with respect to the requirement of anonymizing data will hamper research.

Given the concerns, it is not advisable to completely exempt "anonymous" genetic information from data protection regimes. However, this does not mean that full informed consent for all uses of "anonymous" samples must always be obtained.[75] Other options include security provisions, or practices similar to Statistics Canada for handling aggregate information.[76] Research Ethics Boards will have to pay particular attention to the issues raised by using stored genetic material in research.

The amount of information potentially available from a DNA sample as well as the longevity of the sample raises other concerns regarding the uses of samples and informed consent. For example:

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[73] *Id*., article 10.2 and 10.3.

[74] *Id.* art. 8.6; and discussion at 8.7-8.8.

[75] There is considerable disagreement on the issue of consent in the context of the use of "anonymous" samples in research. See *e.g.*, E.W. Clayton, "Prospective Uses of DNA Samples for Research" in Knoppers *et al*, *supra* note 59, 291at 293-4. She notes that The American Society of Human Genetics takes the position that consent is not required for such use whereas The American College of Medical Genetics recommends consent.

[76] J. Hagey, "Privacy and Confidentiality Practices for Research with Health Information in Canada" (1997) 25 J. L., Med. & Ethics 130 at 132-34.

- When is consent required for the re-testing of stored samples?
- How should individuals provide informed consent to future re-testing of stored samples?
- If information becomes available through future tests on a sample, in what circumstances, if any, should the individual be re-contacted?
- Is it always appropriate for researchers to disclaim responsibility for on-going disclosure of genetic information by obtaining initial informed consent from research subjects in which they accept this, without informing them about potential uses of their DNA sample.[77]
- What if new research provides new information on old test results?
- What should be done regarding secondary information that is gathered as a result of genetic testing or research?
- Should there be any restrictions placed on the uses made of stored samples from deceased individuals?
- If DNA samples are stored for non-research purposes, such as in forensic or military databases, can researchers have access to these samples? If so, under what circumstances?
- Could new genetic findings justify screening of stored DNA for specific health care purposes?

The fact that DNA can be used to uniquely identify an individual also raises concerns regarding the uses that could potentially be made of stored samples. For example, the federal *DNA Identification Act* (not currently in force) puts in place a framework, including safeguards, for the establishment of a national databank containing a crime scene index and a convicted offenders index in order to facilitate the identification of persons alleged to have committed certain designated offences.[78] However, as we have already discussed, DNA databanks are being set up outside of the forensic context as well, and contain genetic information on individuals who have never been suspected of an offence, let alone convicted of one. These may not be research databanks, either. We mentioned already the “Guthrie cards” containing blood spots obtained from newborns for the purpose of screening for PKU and other disorders. DNA is relatively stable in dried blood and so such samples provide a de

[77] For an detailed legal discussion of limitations on research subjects' access to genetic research results, see T. Banks, "Misusing Informed Consent: A Critique of Limitations on Research Subjects' Access to Genetic Research Results." 63 Sask. L. Rev. 539.

[78] S.C. 1998, c.37 [not in force].

facto DNA bank.[79] Nonetheless, the police could gain access to this information through seeking a warrant. This highlights a need to examine the warrant procedures to see if specific guidelines regarding genetic information are required, including regulations regarding the uses of this information once obtained. For example, should the police be allowed access to genetic information that they would not be allowed to obtain under the *DNA Identification Act*? Under what circumstances will the police be allowed to retain the information, for how long, and with what safeguards? These issues are currently dealt with by ss. 487.05 to 487.09 of the *Criminal Code*.[80] However, these provisions address the conditions under which a warrant may be issued to obtain a bodily sample from a particular individual and do not seem to address the situation where what is sought is an already existing sample.[81]

4.3 Uses of Genetic Information

The increasing prevalence of genetic databanks raises questions not only regarding genetic information but also about the *combination* of genetic information and information technology. This issue has particularly come to light with respect to the Human Genome Project. Gene sequencing on such a large scale requires information technology. Not surprisingly, for computer giant Bill Gates, DNA research and computers seem a natural fit.[82] As Dr. Karp, a leader in computer science who works on sequencing projects at the University of Washington, has stated:

> *There's a revolution occurring in biology, particularly at the molecular level. It's turning biology into an information science. Many biologists consider the acquisition of sequences to be boring. But from a computer science point of view, these are first-rate and challenging algorithmic questions.*[83]

The combination of information technology and genetic research makes possible types of research that may raise unique issues, particularly with respect to informed consent.

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[79] J. McEwan, "DNA Databanks" in Rothstein, *supra* note 12, 231 at 245.

[80] R.S., c. C-34.

[81] For example, s. 487.06(1) provides: **"**The warrant authorizes a peace officer or another person under the direction of a peace officer to obtain and seize a bodily substance from the person by means of

(*a*) the plucking of individual hairs from the person, including the root sheath;

(*b*) the taking of buccal swabs by swabbing the lips, tongue and inside cheeks of the mouth to collect epithelial cells; or

(*c*) the taking of blood by pricking the skin surface with a sterile lancet."

[82] See D.L. Wiesenthal & N.I. Wiener, "Privacy and the Human Genome Project" (1996) 6(3) Ethics & Behavior 189 at 193.

[83] Cited in G. Kolata, "Biology's Big Project Turns Into Challenge for Computer Experts" *New York Times* (11 June 1996) C1.

One such issue arises from the possibility of combinatorial rather than hypothesis-driven research. In hypothesis-driven research, researchers note that a particular family, or community, experiences a high incidence of a particular disease. This leads to the hypothesis that the disease has a genetic basis and leads to the search for the particular gene mutation responsible for that disease. Combinatorial research, by contrast, makes use of information technology to compare medical records, genealogical records, and gene sequences in order to discover correlations that may indicate the genetic basis for particular diseases. Such research does not require particular hypotheses to test, but rather relies upon brute computational power to discover genetic links to disease. For example, this is said to be the revolutionary potential of the Icelandic database.[84] But this type of research raises the question of whether individuals should be allowed to give broad consent for the use of their genetic information, as well as their other health information. Broad consent would mean consent to future unidentified uses of their information, and the question is whether such consent can be meaningful.

As we have already discussed, genetic information also relates to one's biological relatives. This reason makes families particularly important for genetic research, but increasingly whole populations such as in Iceland, or Newfoundland, or various aboriginal communities, are of interest to researchers. As the kind of whole population study such as in the Icelandic database becomes more prevalent, made possible by the combination of genetic research and information technology, then community concerns need to be taken into account. [85] The following are some issues that need to be dealt with:

- Do researchers need to get informed consent from an entire community, or simply individual participants?
- What are the acceptable mechanisms through which to obtain informed consent from a community?
- Should a community receive any benefits for agreeing to participate in genetic studies?
- If a community is offered benefits to participation, then should safeguards be put into place to ensure that this does not become coercive?

[84] On these issues, compare G.J. Annas, "Rules for Research on Human Genetic Variation – Lessons from Iceland" (2000) 342 New England Journal of Medicine 1830 and J.R.G. & K. Stefansson, "The Icelandic Healthcare Database and Informed Consent" (2000) 342 New England Journal of Medicine 1827.

[85] S.P. Hoffert, "Concerns Mount over Privacy as Genetic Research Advances" (1998) 12(2) Scientist 1.

- How can people who opt out be protected from the negative impact of receiving unwanted information that also may pertain to them?

Finally, the drive to identify the genetic basis of disease often has a commercial aspect, particularly when researchers seek patents on the genetic sequences they identify, and the diagnostic tests they develop on the basis of these sequences. While gene patenting is controversial for many reasons beyond the scope of this discussion, one issue that is of relevance here is informed consent. To what extent and how do patients and research subjects have to be informed of the potential commercial applications of research? The Tri-Council Policy Statement explicitly imposes a duty on researchers to discuss this with research subjects.[86] A recent controversy around the patenting of the Canavan disease highlights how sensitive these issues are. In this case, parents of children who suffered and died from Canavan disease sued researchers involved in the research and subsequent patenting of the gene. Family members of children with Canavan-disease were particularly upset because those who contributed to the discovery of the gene now had to pay for testing of other family members.[87]

[86] Social Sciences and Humanities Research Council, Natural Sciences and Engineering Research Council, Medical Research Council, *supra* note 72, art. 8.7.

[87] P. Gorner, "Parents Suing over Patenting of Genetic Test" *Chicago Tribune* (November 19, 2000).

5. CONCLUSION

In this paper, we have indicated that different characteristics attributed to genetics do not necessarily make genetic information something entirely new and unique. However, the combination of some of these characteristics, and the way some of them play out in the context of genetics, do warrant a careful assessment of existing regulatory approaches towards health information. These characteristics, combined with the particular volume of data, the potential speed of testing and the link with computer technology, make it necessary to investigate how we can better protect private health information. More comprehensive protective measures are warranted. One of us has argued elsewhere that it may be inappropriate to single out genetics in specific genetic anti-discrimination legislation, particularly in the context of insurance.[88] However, it was also stressed that we need to develop flexible regulatory structures to analyse how developments in genetics impact on society and to intervene when protection is needed. In many cases, existing regulatory regimes could be adapted, for example to make sure that regulatory definitions capture new genetic information. As we stressed in our discussion, when developing new regulations, attention will have to be paid to the various ways in which genetic information is gathered, circulated and used. In the coming years, additional efforts will have to be made to analyse carefully whether existing regulations and laws capture all the issues raised by genetic technologies, how we can adapt them, and what new initiatives are needed. Computer technology has accelerated genetic research. Social concerns about some consequences of genetic research will hopefully accelerate the pace of regulatory change aimed at improving the protection of individuals and communities against informational and other harms.

[88] "Selective Justice", supra note 15, in particular at 407-412.

GENETICS, PRIVACY AND DISCRIMINATION

Document Prepared for the Canadian Biotechnology Advisory Committee

Project Steering Committee on Genetic Privacy

By

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EXECUTIVE SUMMARY

Genetic information about individuals – personal genetic information – has many current and potential uses: to assist in predicting, diagnosing, treating and preventing health conditions; to assist with reproductive decisions; to decide suitability for employment; to assess the health consequences of exposure to workplace contaminants – for example, radiation; to assess eligibility for services such as insurance and credit; as an identification tool in criminal investigations; to advance medical research; to verify gender in sports competitions; to determine paternity; and to assess the susceptibility of ethnic groups to genetically-tuned biological weapons.

The relatively recent advent of "molecular genetics" has increased the number of physical and behavioural characteristics that may be revealed by genetic testing. This in turn has heightened privacy concerns and fears of discrimination based on genetic "makeup." That discrimination could take several forms – rejection for employment, restricted access to credit or insurance, and even discriminatory treatment in government programs dealing with reproduction and education.

Western countries generally have enacted little legislation dealing *specifically* with genetic privacy and discrimination. However, the list of laws and proposed laws applying specifically to genetics is growing, particularly in the United States and mainly in three areas – insurance, employment and criminal forensics.

In Canada, apart from criminal legislation on using DNA in criminal investigations, most provisions affecting genetic privacy and discrimination are found in laws of more general application. They appear in constitutional law, laws governing professional confidentiality, an emerging set of provincial laws dealing with health information, data protection (privacy) and human rights laws, statutory torts, and the criminal law (protections against physical intrusions). Many of these general laws were drafted without genetics in mind. Still, they provide a substantial, if incomplete, legal framework for handling personal genetic information.

Attempts to protect genetic privacy have also been made at the international level, such as through the 1997 European *Convention* on *Human Rights and Biomedicine* and the *Universal Declaration on the Human Genome and Human Rights* (UNESCO, 1997). Other more general international instruments are also relevant. These include research guidelines and conventions dealing with human rights generally.

General Issues

Tension Between the Potential Benefits and Harms of Genetic Technology: At present, many benefits of genetic science remain theoretical. However, the misuse of genetic information about individuals has already led to genetic discrimination, sometimes about overt genetic characteristics such as skin colour or gender, and other times about genetic traits discoverable only through testing – sickle cell anemia, for example.

Is Genetic Information an Exceptional Form of Personal Information? Debate continues about whether genetic information is somehow "exceptional", requiring different, perhaps more cautious and protective treatment than other types of personal information.

The Right Not to Know: Respect for individual autonomy can be used as the basis to argue that individuals should not be forced to acquire genetic information about themselves. Such knowledge could be catastrophic – such as learning, against one's wishes, that one has the gene that causes Huntington disease. As well, there is debate whether minors have or should have an equivalent right not to know, or whether their guardians should be permitted to obtain information that the minors themselves might not later want?

Secret and Private Testing: Individuals may soon be able to identify a number of genetic traits through commercially available testing kits. These kits will inevitably invite the surreptitious testing of others. Even if not used surreptitiously, the very availability of these kits to the general public may encourage misuse – for example, to defraud insurance companies.

Disclosure to Biological Relatives: Test results about a person may identify genetic traits of biological relatives. There is considerable debate about whether a duty or ethical obligation exists on professionals or individuals to share useful genetic information with biological relatives.

Discrimination on the Basis of Perceived Disability: Case law and legislation have extended the protection against discrimination on the basis of disability to cases of perceived disability. Thus, the potential for discrimination on the basis of perceived disability is significantly reduced. However, the extent to which human rights legislation protects against discrimination because of a possible *future* genetically linked disability remains unclear.

A Residual Right of Genetic Privacy? Even if legislation, codes, ethical standards and other instruments were to provide generous confidentiality protection, some argue that there is nonetheless a residual right to say "no" to further uses of one's genetic information. This issue is most germane in the context of research.

Specific Areas of Concern

Human Reproduction: Governments will inevitably be drawn to programs that prevent the birth of children with expensive genetic "disabilities." Subsidiary issues also arise, among them how to prevent further dissemination of genetic information acquired by private reproductive clinics, and rights, if any, of children conceived as a result of a sperm or egg donation to learn the identity, or at least the genetic background, of the donor, and the potentially conflicting rights of the donor to confidentiality.

Employment: Employers may want genetic information about employees or job applicants. If the burden of health care costs shifts to the private sector, Canadian employers may become even more interested in hiring only the healthiest employees.

Testing to Determine Eligibility for Services Such as Insurance and Credit: Genetic information may further separate those who have access to insurance, credit and other services from those who, because of their genetic makeup, do not.

Conclusions

The possible use of personal genetic information against individuals may justifiably stifle acceptance of further genetic inquiry. Failure to protect privacy and prevent discrimination therefore risks greatly diminishing the potential for genetics to improve health care.

The key to benefitting from genetic information while avoiding its drawbacks lies in controlling use of the information beyond the health care of the individual to whom the information relates. Regulation and, in some cases, prohibitions, on secondary uses of personal information are indispensable once personal genetic information has been collected. DNA collected and analyzed for health care purposes should not automatically be available for further uses, even research, if the DNA can be linked to an identifiable individual.

The ultimate protection, however, may often lie in more strictly limiting the *initial* collection of personal genetic information. For example, the greatest protection against state interference with human reproduction will come from keeping personal genetic information from the state in the first place.

General laws governing personal health information can often protect genetic information, although sometimes these general laws themselves are inadequate. However, genetic information brings new intensity to the need to protect personal health information. Legislation to address specific issues relating to genetic testing maybe required to supplement existing legislation. Legislation dealing with the taking of DNA from criminal suspects and the establishment of DNA databanks relating to convicted offenders must be carefully monitored to prevent an unwarranted enlargement of its scope.

Public education is essential to protect genetic privacy and prevent discrimination. Governments in particular have a duty to explain the uses of genetic information and their possible impact on society.

INTRODUCTION

Advances in genetic science give rise to many concerns – among them the violations of privacy inherent in collecting and analyzing genetic material. The possible further consequence of these violations, and the consequence that many fear most, is genetic discrimination – discrimination on the basis of one's genetic "makeup."

Discrimination may take any number of forms – rejection for employment, or the offer of lesser employment, loss of access to credit or insurance, or access only under extraordinary conditions and at extraordinary expense, and even discriminatory treatment in the application of government social policies relating to reproduction and education. The unwanted collection and release of genetic information may also interfere with personal relationships. A potential marriage partner may reject someone with a genetic risk of contributing to a "defective" child.

Above all, the promise of genetics for improved health and health care may be severely compromised unless privacy and discrimination issues are addressed. For example, a 1998 survey conducted for the US National Center for Genome Resources found that almost two-thirds of the respondents said they probably or definitely would not take genetic tests if health insurers or employers could get access to the results.[1] The possibility that genetic information will be used to the disadvantage of individuals, rather than to help them, may – justifiably – stifle acceptance of further genetic inquiry at a time when a major milestone in understanding genetics – the initial sequencing of the human genome – has just been reached. This analysis is structured as follows:

Part I provides a cursory analysis of the relevant science relating to genetics and privacy.

Part II provides an overview of the legislative schemes in Canada dealing with privacy and discrimination generally. Part II also outlines specific initiatives aimed at genetics issues, both in Canada and in several other jurisdictions.

[1] "Employers Should Be Barred From Accessing Genetic Information, Americans Say In NCGR Survey," March 4, 1998: http://www.ncgr.org/about/news/archive.html.

Part III explores a broad range of genetic privacy and discrimination issues.

Part IV contains recommendations for specific action to protect genetic privacy and prevent discrimination.

The range of genetic privacy and discrimination issues is enormous and growing. This brief paper can only touch on some of the major themes. It cannot fully address the vastly complex issues of genetics, privacy and discrimination – issues that for more than a decade have dominated much of the ethical and legal debate surrounding genetics. For reasons of brevity, it does not deal in any detail with forensic applications of DNA in criminal investigations or the use of genetic information to enhance the targetting of biological weapons.

PART I: THE SCIENCE

1. WHAT GENETIC TECHNOLOGY CAN REVEAL

Genetic technology is not new. Simple tests have been used for decades to identify chromosomal problems.[2] However, the relatively recent advent of "molecular genetics", which enables the identification of genetic defects in the DNA molecule itself, has magnified the impact of genetic testing. As one author states, "the ability to identify genetic defects in the DNA molecule itself has led to a higher degree of specification of genetic disorders than has ever before been possible."[3] This enhanced degree of detail about behavioural and physical characteristics has intensified the privacy and discrimination issues relating to personal information generally.

The extent to which individual characteristics and behaviours are determined by genes is the focus of the debate surrounding "genetic determinism" – the belief that all human behaviour is governed by a chain of determinants that runs from the gene to the individual to the sum of the behaviours of all individuals.[4]

Many claims about genetic links to diseases or behaviours are tentative. Some generate great controversy. Some researchers, for example, argue that genes appear to contribute to homosexuality; others dismiss this link.[5] Amidst this still unresolved debate about the impact of genetics on characteristics and behaviours, "discoveries" about genetic links to diseases or behaviours occur with increasing frequency. Among the many genetic "discoveries" of the past few years, for example, have been a "salt gene" that could explain why some people with high blood pressure respond to a low-salt diet, and others do not; a finding that black

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2 J.T.R. Clarke, "Professional Norms in the Practice of Medical Genetics", [1995] 3 Health Law Journal 130.

3 Clarke, above, at 138 .

4 Cited in Bartha Knoppers, Human Dignity and Genetic Heritage, a Study Paper prepared for the Law Reform Commission of Canada (Ottawa: Law Reform Commission of Canada, 1991) at 43.

5 L. Hood and L. Rowen, "Genes, Genomes, and Society", in Mark Rothstein, ed., *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (New Haven, Yale University Press, 1997) at 27; but see *The Independent*, May 3, 1998: "Despite claims that area Xq28 of the X chromosome contains a gene giving a "tendency" to homosexuality, scientists dismiss the idea."

smokers appear to absorb more nicotine than white smokers, which could explain why black smokers have more trouble quitting and run a higher risk of lung cancer; a report that people who are miserable and depressed may have been born with a genetic predisposition not to be happy; a discovery about several genetic defects that increase the tendency to put on weight; a report about a genetic mutation that can cause heart failure; a report that a "novelty-seeking" gene may influence sensation-seeking in adults; a finding that one gene plays a key role in inflammatory breast cancer, the most deadly form of the disease; a finding that even dark-skinned people who carry certain genetic variations are at increased risk for skin cancer; and the discovery that alteration of a specific gene appears to contribute to both the common late-onset form of Parkinson's disease, and the rarer, early-onset form of the disease.

2. USES OF GENETIC INFORMATION

Genetic information can be useful in several situations, among them:

- to assist in predicting, diagnosing, treating and preventing health conditions;
- to assist in making reproductive choices and decisions relating to reproduction generally;
- to assess suitability for employment;
- to assess the genetic consequences of exposure to certain workplace or environmental materials or contaminants – for example, radiation;
- to assess eligibility for services such as insurance and credit;
- as an identification tool in criminal investigations;
- for medical research;
- to verify gender in sports competitions;
- to determine paternity; and
- to assess the susceptibility of ethnic groups to genetically-tuned biological weapons.

Most genetic diseases involve many genes and often also involve environmental components (that is, they are "multifactorial" diseases). Such diseases include hypertension, diabetes and coronary heart disease. Multifactorial diseases are highly difficult to predict through genetic testing simply because so many genes and environmental factors may be involved. Scientists may be able to say little more than that the presence of a particular gene or genes contributes to the risk of acquiring a disease, but they cannot state with certainty whether the individual will develop the disease. Contrast this with the relatively rare single gene (monogenic) diseases, where a mutation in a single gene can indicate a certainty of acquiring the disease.

PART II: LEGISLATIVE SCHEMES DEALING WITH GENETIC PRIVACY AND DISCRIMINATION

1. THE GENERAL LEGAL FRAMEWORK

In many Western countries, there appears to be little legislation dealing *specifically* with genetic privacy and discrimination. However, the list of laws and proposed laws applying specifically to genetics is growing, particularly in the United States and primarily in three areas – insurance, employment and criminal forensics.

In Canada, apart from legislation dealing with the use of DNA in criminal investigations, most provisions relevant to genetic privacy and discrimination are not found in laws dealing specifically with genetics issues. Instead, they appear in more general legislation – constitutional law, laws governing professional confidentiality, data protection (privacy) and human rights laws among them. Many of these general laws were drafted without genetics in mind. Nonetheless, they provide a substantial, if incomplete, legal framework for handling personal genetic information.

a. Constitutional Law

Some protection of personal genetic information from misuse by government is found in the *Charter of Rights*. This protection occurs through the fundamental rights of freedom of association, conscience and religion; life, liberty and security of the person; freedom from unreasonable search and seizure; and the right to equality.[6] The *Charter* is clearly an important vehicle for protecting genetic privacy and avoiding genetic discrimination at the hands of government.

[6] Knoppers, above, at 41-42.

These *Charter* rights, however, are not absolute and must be read in light of section 1 of the *Charter*, which can limit the rights stated elsewhere in the *Charter*. Even so, stringent conditions must be met before section 1 limits a constitutionally protected right.

b. Federal and Provincial Data Protection Laws

Parliament and most provincial legislatures have now enacted data protection legislation that regulates the collection, use and disclosure of personal information by governments and many government agencies. Until recently, only Quebec had enacted data protection legislation regulating the private sector. As of January 2001 federal legislation, the *Personal Information Protection and Electronic Documents Protection Act*,[7] will regulate the collection, use and disclosure of personal information by commercial organizations that are federally regulated. It also provides individuals with a right of access to information held about them. However, the Act exempts "personal health information" for one year after the legislation comes into force. The collection, use or disclosure of personal health information will not be covered until January 2002.[8] Personal health information includes "information derived from the testing or examination of a body part or bodily substance of the individual,"[9] and therefore includes genetic test results. Confusion remains about whether the Act covers physicians, since the Act applies to commercial organizations, and there is some doubt whether this includes the traditional professions.

If provincial governments fail to enact similar data protection legislation governing provincially regulated commercial activities within three years of the Act coming into force,[10] the *Personal Information Protection and Electronic Documents Protection Act* will extend to all commercial activity, both federal and provincial.

Data protection legislation is clearly relevant in discussing genetic privacy. Genetic information about an individual is "personal information," precisely the type of information that data protection legislation is intended to regulate. However, data protection legislation varies from jurisdiction to jurisdiction and is often not an effective guardian of genetic information. The federal *Privacy Act*,[11] for example, imposes only loose restrictions on the collection and disclosure of personal information by the federal government.

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7 S.C. 2000, c. 5.

8 Sections 30(1.1) and (2).

9 Section 2.

10 Section 30(2).

11 R.S.C. 1985, c. P-21, sections 4 and 8.

c. Provincial Health Information Legislation

Three provinces – Manitoba, Saskatchewan and Alberta[12] – have recently enacted legislation to deal specifically with privacy and confidentiality of health care information, and Ontario is considering such a move. These acts regulate the collection, use and disclosure of medical records, including genetic records. As well, provincial legislation regulating health care and health care institutions often contains provisions protecting the confidentiality of medical information by limiting its further disclosure;[13] such provisions would apply to genetic information.

d. Human Rights Codes

Human rights codes generally prohibit discrimination in employment and access to services on the basis of disability, and case law has extended the protection to cases of perceived disability. In Ontario, the legislation makes it clear that protection extends to cases of perceived disability. Thus, the potential for genetic discrimination is significantly reduced by existing human rights codes and by the decisions interpreting those codes,[14] including a recent Supreme Court of Canada decision.[15] There, the Court emphasized that the right to protection against discrimination on the basis of disability covers discrimination based on *perceived* disability. Still, the extent to which *future* genetic disability is protected by human rights codes is not clear.

There has been some action in the United States to protect employees against genetic discrimination. The American Civil Liberties Union reported in 1998 that 12 states had enacted laws that protect employees from genetic discrimination in the workplace and that a handful of other states had legislation pending at that time.[16] A 1999 compilation of employment laws prepared by the US National Human Genome Research Institute showed

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[12] Manitoba: Personal Health Information Act (1997); Saskatchewan: Health Information Protection Act (1999); Alberta: Health Information Act (1999).

[13] For example, the Nursing Homes Act, R.S.O. 1990, c. N.7, section 6; Homes for the Aged and Rest Homes Act, R.S.O. 1990, c. H.13, section 6; Long-Term Care Act, 1994, S.O. 1994, c. 26, sections 3(1) and 32 (1); Ontario Drug Benefit Act, R.S.O. 1990, c. 0.10, section 13(6).

[14] See generally the discussion in Trudo Lemmens and Poupak Bahamin, "Genetics in Life, Disability and Additional Health Insurance in Canada: A Comparative Legal and Ethical Analysis", in Bartha Knoppers, ed., Socio-Ethical Issues in Human Genetics (Cowansville: Les Éditions Yvon Blais, Inc. 1998) 114 at 201-09.

[15] Quebec (Commission des droits de la personne et des droits de lajeunesse) v. Montréal (City); Quebec (Commission des droits de la personne et des droits de la jeunesse) v. Boisbriand (City) [2000] 1 S.C.R. 665.

[16] "Genetic Discrimination in the Workplace Fact Sheet", (1998) http://www.aclu.org/issues/worker/gdfactsheet.html (April 5, 1999).

that 25 states had to that time enacted provisions on using genetic information in employment.[17] No federal legislation has been passed in the US relating to genetic discrimination in individual insurance coverage or to genetic discrimination in the workplace. However, several federal bills were introduced during the last decade, and on February 8, 2000, President Clinton signed an executive order prohibiting every federal department and agency from using genetic information in any hiring or promotion action.[18]

e. Insurance Law

Human rights legislation does not prevent discrimination in insurance. In fact, current insurance law promotes the use of medical information for underwriting.[19] For example, model provincial insurance legislation, the *Uniform Insurance Act*, requires an applicant for insurance to disclose to the insurer "every fact within the person's knowledge that is material to the insurance . . . ."[20] No exception is made for genetic information.

Contrast this with the United States. A 1998 Associated Press report states that 150 million Americans insured at work have legislative protection against some forms of genetic discrimination in insurance. The report states as well that 24 states restrict what insurers can do with genetic information.[21] A 1999 compilation of US insurance laws prepared by the US National Human Genome Research Institute showed that 41 states had enacted provisions relating to genetic privacy in insurance matters.[22]

In the United Kingdom, there appears to be no legal prohibition against genetic testing in insurance matters. In November 1998, the Department of Trade and Industry announced a voluntary agreement with insurance companies. Among the terms of the agreement: all genetic tests must be individually validated before they can be used by the insurance industry, and those who take genetic tests are to have the right to keep the results from life insurance companies.[23] This right would last only until insurance companies can show that a genetic test has a proven ability to predict a person's premature death.

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17 http://www.nhgri.nih.gov/Policy_and_public_affairs/Legislation/workplace.htm (accessed October 31, 2000).

18 http://www.ornl.gov/hgmis/elsi/legislat.html (accessed October 31, 2000).

19 Lemmens and Bahamin at 271.

20 Lemmens and Bahamin at 190.

21 "Test Patients Fear Losing Insurance", Associated Press, April 11, 1998 (New York).

22 http://www.nhgri.nih.gov/Policy_and_public_affairs/Legislation/insure.htm (accessed October 31, 2000).

23 The Independent, November 14, 1998.

In July 2000, the Association of British Insurers submitted an application to the Genetics and Insurance Committee (GAIC), set up by the UK Department of Health, for approval of two genetic tests for Huntington Disease. In October 2000, the GAIC, announced that the reliability and relevance of the genetic test was sufficient for insurance companies to use the result when assessing applications for life insurance.[24]

f. Statutory Torts

Four provinces – British Columbia,[25] Saskatchewan,[26] Manitoba[27] and Newfoundland[28] – have enacted statutory privacy "torts." These laws make it a civil wrong to violate the privacy of another person without justification. In Quebec, there is no statutory tort of violation of privacy. However, article 1053 of the Quebec *Civil Code* may provide similar protection.

g. Professional Codes

Provincial legislation governs the professional conduct of physicians and some other health care professionals. Often, however, the legislation does not explicitly set out a duty of confidentiality as part of its standards for professionals. This gap is sometimes closed by relying on the confidentiality provisions of codes of professional conduct.[29] One voluntary code, the Canadian Medical Association's *Health Information Privacy Code*,[30] sets out the minimum requirements to protect the privacy of patients and the security and confidentiality of their health information. The code covers the collection, use, and disclosure of personal health information and rights of access to the information.

h. Laws Protecting Against Physical Intrusion

The criminal law may prevent the forced taking of DNA samples on which to do genetic testing. The physical intrusions necessary to obtain saliva, blood or hair could amount to a

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[24] "Insurance in the genetic age," The Economist, October 21, 2000.

[25] R.S.B.C. 1979, c. 336, section 1(1).

[26] The Privacy Act, R.S.S., c. P-24, section 2.

[27] The Privacy Act, C.C.S.M., P125.

[28] The Privacy Act, S.N. 1981, c. 6, section 3(1).

[29] Gilbert Sharpe, The Law and Medicine in Canada, 2d ed. (Toronto: Butterworths, 1987) at 223-24. Though the text is already dated, the commentary appears to remain relevant

[30] Approved by the CMA Board of Directors, August 15, 1998.

criminal assault if they occurred without the consent of the person or without specific legislative authority[31] to take the samples. This is so even if the physical intrusion itself is very minor. Obtaining DNA without consent could also constitute civil battery.

i. Possible Quasi-Constitutional Protection: the Privacy Rights Charter (Bill S-27)

Because Canadian constitutional law lacks an explicit constitutional right to privacy, the Hon. Sheila Finestone introduced a private senator's bill, Bill S-27, the *Privacy Rights Charter*, on June 15, 2000. The bill, intended to give privacy quasi-constitutional status, would guarantee the right of the individual to privacy. It would define what is an infringement and provide a test for justifiable infringement. It would also entitle individuals to claim and enforce their right to privacy, and to refuse to unjustifiably infringe the privacy rights of others. It would prohibit unjustifiably infringing the right to privacy of another individual.

The bill would also require the Minister of Justice to review bills and regulations for compliance, and entitles the Privacy Commissioner of Canada to be consulted in this regard.

Unlike the *Charter of Rights*, which applies only to government, the *Privacy Rights Charter* would also apply to the federally-regulated private sector and could serve as a template for similar provincial legislation. (Only Quebec, with its *Charter of Human Rights and Freedoms* has already give a similar quasi-constitutional status to privacy, by affording every person the right to respect for his or her private life.)

Bill S-27 died when the federal election was called in October 2000, but will very likely be re-introduced after the election.

[31] Such as that provided by 1995 and 1998 amendments to the Criminal Code (and by parallel amendments to the National Defence Act that came into effect on June 30, 2000) to permit the police to take DNA samples from certain criminal suspects and from those convicted of certain criminal offences.

2. EXAMPLES OF CANADIAN LAWS AND OTHER INITIATIVES DIRECTED SPECIFICALLY AT GENETICS ISSUES

a. Bill C-47: Human Reproductive and Genetic Technologies Act

Bill C-47, the *Human Reproductive and Genetic Technologies Act*,[32] was introduced in the House of Commons in 1996. The Bill sought to prohibit the use of certain reproductive and genetic technologies (including cloning) in relation to human beings, as well as certain commercial arrangements relating to human reproduction. The bill would have prohibited performing any medical procedure to ensure or increase the probability that a zygote or embryo will be of a particular sex, except for reasons related to the health of the zygote or embryo.[33] As well, the bill would prohibit performing any diagnostic procedure to ascertain the sex of a zygote, embryo or fetus, except for reasons related to its health.[34]

Bill C-47 died on the Order Paper. The Minister of Health then announced that he would introduce comprehensive legislation – a combination of prohibitions and a regulatory regime – before the end of 1999. When the federal election was called in October 2000, no such legislation had yet been introduced.

b. Research Ethics

The August 1998 Tri-Council Policy Statement,[35] *Ethical Conduct for Research Involving Humans*, contains several provisions relating to privacy and genetic research involving human subjects. The policy statement does not have the force of law, but it offers strong guidance on ethical issues relating to genetic privacy and discrimination. Among the issues covered by the guidelines are the potential loss of benefits and other harms flowing from the further use of genetic information, commercial uses of genetic information and the banking of DNA material.

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[32] 2nd Sess., 35th Parl., 1996-97.

[33] Clause 4(1)(h).

[34] Clause 4(1)(i).

[35] Medical Research Council of Canada, Natural Sciences and Engineering Research Council of Canada, Social Sciences and Humanities Research Council of Canada, *Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans* (August 1998).

c. Report of the Privacy Commissioner of Canada, Genetic Testing and Privacy

In 1992, the Privacy Commissioner of Canada issued *Genetic Testing and Privacy*.[36] The report examined genetic privacy issues relating to employment, access to services, human reproduction and criminal investigations, among other topics. The report spoke of the need to protect the privacy and confidentiality of personal genetic information, and made 22 recommendations for that purpose. Among other topics, these recommendations dealt with the right "not to know," the need to restrict or prohibit collection of genetic information by governments, employers and service providers, and the use of DNA in criminal investigations.

d. The Canadian Genome Analysis and Technology Program (CGAT)

The Canadian Genome Analysis and Technology Program was established in 1992 as the Canadian arm of the International Human Genome Project. CGAT established an advisory committee on research into the medical, ethical, legal and social implications ("MELSI") of genetics in 1993. The MELSI advisory committee sought to identify priority medical, ethical, legal and social issues in Canada. CGAT funded numerous studies into these issues. Among the issues addressed by the advisory committee were genetics and insurance, and comparative international approaches to genetics issues. This work, as a component of the larger CGAT, ended in April 1997.

e. House of Commons Standing Committee on Human Rights and the Status of Persons with Disabilities

The Standing Committee made several recommendations relating to genetic discrimination in its April 1997 report on privacy rights and new technologies, entitled *Privacy: Where Do We Draw the Line?* The Committee called for immediate action to deal with privacy violations and discrimination flowing from genetic testing. It called for a review of genetic testing policies and practices in several areas -- employment, health, insurance, and criminal justice.

36 (Ottawa: Minister of Supply and Services Canada, 1992).

3. INTERNATIONAL INITIATIVES

a. The Proposed Genetic Privacy Act

In 1995, model legislation, the *Genetic Privacy Act*, was completed at the Health Law Department, Boston University School of Public Health.[37]

The authors described the Act as "a proposal for federal [U.S.] legislation. The Act is based on the premise that genetic information is different from other types of personal information in ways that require special protection." The model act would impose tight restrictions on the collection, use and disclosure of genetic information. As well, there would be special rules regarding the collection of DNA samples for genetic analysis for minors, incompetent persons, pregnant women, and embryos. Exceptions would be made for DNA samples collected and analyzed for identification for law enforcement purposes if authorized by state law, and for identifying dead bodies. Research on individually identifiable DNA samples would be prohibited unless the individual authorized the research use, and research on nonidentifiable samples would be permitted if not prohibited by the individual.[38]

b. Council of Europe Convention on Human Rights and Biomedicine 1997

The 1997 European *Convention on Human Rights and Biomedicine*[39] was opened for signature by Council of Europe member states, and non-member states which participated in its development, including Canada. As of October 2000, the treaty had not received sufficient ratifications from Council of Europe member states for it to enter in force. As well, Canada had not yet signed or ratified the treaty.

The convention bans all forms of discrimination based on the grounds of a person's genetic make-up and allows the carrying out of predictive genetic tests only for medical purposes. The Convention also sets out rules for medical research and recognizes a patient's right "not to know."

[37] Source: http://www.ornl.gov/TechResources/Human_Genome/resource/privacy/privacy1.html (April 2, 1999).

[38] http://www.ornl.gov/TechResources/Human_Genome/resource/privacy/privacy1.html#intro (April 2, 1999).

[39] ETS No. 164.

c. Universal Declaration on the Human Genome and Human Rights (UNESCO 1997)

In 1997, the General Conference[40] of UNESCO adopted the *Universal Declaration on the Human Genome and Human Rights.* The Declaration contains several provisions aimed at preventing genetic discrimination – for example, the right of everyone to respect for their dignity and human rights regardless of their genetic characteristics. Furthermore, "[t]hat dignity makes it imperative not to reduce individuals to their genetic characteristics and to respect their uniqueness and diversity." The Declaration also prohibits discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity. It also proposes strict rules for genetic research.

d. Other International Instruments

Even if not specifically directed at genetics issues, many international instruments have a bearing on genetic privacy and discrimination. These include declarations and conventions dealing with human rights generally, research guidelines and conventions dealing with matters such as biological weapons.

Declarations and conventions on human rights, for example, address privacy rights, equality rights, rights of association, freedom of religion, the right to adequate health care and the right to establish a family. International agreements or guidelines on research may also help prevent genetic discrimination. Prohibitions against the development and use of biological weapons are also directly relevant to genetic discrimination, particularly as evidence mounts that genetic characteristics are being studied as possible ways to help target biological weapons against specific ethnic groups.[41]

[40] 29th Session.

[41] British Medical Association, *Biotechnology, Weapons and Humanity*, Harwood Academic Publishers, 1999.

PART III: THE ISSUES

1. OVERRIDING ISSUES

a. Tension Between the Potential Benefits and Harms of Genetic Technology

Genetic technology shows great potential for the prediction, diagnosis, treatment and prevention of disease. At present, many of these benefits remain potential, since science has only an incomplete understanding of the complexities of health, disease and genes.

Threats to privacy and the risk of discrimination, however, are not merely theoretical. The use and misuse of genetic information about individuals has already led to genetic discrimination, sometimes about overt genetic characteristics such as skin colour or gender, and other times about genetic traits discoverable only through testing – sickle cell anemia, for example. Genetic information will almost certainly continue to result in discrimination and other violations of human rights unless individuals retain control over such information.

b. Is Genetic Information an Exceptional Form of Personal Information?

Is genetic information somehow "exceptional," requiring different, perhaps more cautious and protective treatment than other types of personal information? This issue has not been resolved.

Some genetic information, such as eye colour, is normally benign; other such information promises to reveal secrets that may profoundly affect the lives of individuals and their families. It may simply be that genetic information brings privacy and discrimination issues into sharper relief than do some other types of personal information. Thus, genetic information may add urgency and depth to issues that have long been present.

c. The Right Not to Know

Individuals are not normally obliged to investigate their health status. This right not to know can be viewed as an aspect of individual autonomy.

Respect for autonomy can be used to support the argument that individuals should not be forced to acquire genetic information about themselves. In many cases, such information might not harm the individual. In other cases, however, this knowledge could be catastrophic – such as learning, against one's wishes, that one has the gene that causes Huntington disease.

The Privacy Commissioner of Canada has argued that everyone has a right to a reasonable expectation of genetic privacy. This includes the right "not to know" about oneself.[42]

Another aspect of this issue relates to minors. Should a minor have an equivalent right not to know? If a genetic test is looking for a condition for which no treatment is now available, or if a test is predictive of a disorder that will occur only in adulthood, the moral and ethical case for parents to seek genetic testing for a minor may be weak, even if the parents have the right in law to seek the testing. A recently reported technique to test every chromosome in a human embryo before it is implanted in the womb raises equally troubling issues. A news report about the technique states that it will make it possible to know the entire genetic makeup of a baby before it is born.[43] If so, the child could be born already saddled with a genetic "pedigree" that he or she may not later want to know as an adult.

d. Secret and Private Testing

An emerging factor in the debate over genetics, privacy and discrimination is the prospect of widely available private genetic tests.[44] As now occurs with home pregnancy test kits, individuals may one day be able to identify specific genetic characteristics by using a commercially available, and likely increasingly affordable, test. Similarly, they could use such tests to determine the genetic characteristics of anyone whose genetic material – saliva or hair roots, for example – is accessible to them.

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[42] *Genetic Testing and Privacy*, above, at 30-31.

[43] "Genetic test opens door to quest for 'perfect babies,'" *Ottawa Citizen*, October 23, 2000.

[44] See, for example, "Private gene testing should be allowed on trial basis -- bioethicist," Canadian Press September 13, 1998, 23.03 EST (Edmonton).

The main impact of the tests lies in the likely expansion of situations where people will be tested, perhaps even without their knowledge. Easier commercial availability of testing kits will inevitably invite the surreptitious testing of others. Even if not used surreptitiously, the availability of these diagnostic tools to the general public may encourage their misuse.

The commercial availability of private genetic tests also has implications for the insurance industry. Those with a family history of a debilitating disease might use such a test to determine if they are at risk for the disease. If they are, they might then buy a large amount of life or disability insurance, but (fraudulently) without telling the insurance company of their increased risk. This "adverse selection" could impose an unfair burden on the insurance industry.

e. Disclosure to Biological Relatives

There is considerable debate about whether a duty exists to share useful genetic information with biological relatives. Such information may safeguard the health or lives of those relatives. However, disclosing that information reveals the genetic traits of the family member who was tested, and that person may not want the test results disclosed to other family members. This poses serious ethical and legal dilemmas for healthcare workers who hold this information. Should they breach the obligation of confidentiality they owe their patient in order to protect the lives or health of biological relatives, or should the patient's right to confidentiality prevail?

f. Discrimination on the Basis of Perceived Disability

Case law has extended the protection against discrimination on the basis of disability to cover perceived disability, and the Ontario human rights legislation offers explicit protection against such discrimination. Thus, the potential for discrimination on the basis of both actual and perceived genetic disability is significantly reduced.

However, the extent to which human rights legislation and case law protect against discrimination due to the possibility of a *future* disability remains unclear. If an employer refuses to hire a person because the person has a genetic trait that may or will lead to disease, but the employer still considers the person to be healthy now, does that amount to discrimination on the basis of disability or perceived disability?

g. A Residual Right of Genetic Privacy?

Even if legislation, codes, ethical standards and other instruments were to provide adequate confidentiality protection, is there nonetheless a residual right to say "no" to uses of one's genetic information beyond the originally intended use?

This question is most germane in the context of research. Should a person with a severe genetic disability have the right to refuse researchers access to his or her genetic information, even if that research may be in the public interest? What if the goal of the research is to locate the gene that causes or contributes to the disability, so that in future it may be possible to "screen out" that disability from the population? Should a person have a right to refuse to participate in research if the ultimate goal of the research is to prevent the birth of others like that person?

Furthermore, even if there is no ethical issue such as that identified above, is there a residual right to be arbitrary – to deny someone else access to personal genetic information for no reason other than to assert one's right to control personal information about oneself?

2. ISSUES RELATING TO SPECIFIC SITUATIONS

a. Human Reproduction

Some of the most troubling privacy and discrimination issues relate to human reproduction. Cost-conscious governments will inevitably be drawn to programs that prevent the birth of children with genetic "disabilities." Governmental pressure could take several forms:

- relatively neutral advice to prospective parents about the risk of giving birth to a genetically "defective" child;
- advice to parents not to have children, or to abort a "defective" fetus;
- positive financial incentives to abort or not to conceive;
- imposition of financial responsibility for the additional health care and other costs arising from the birth of a "defective" child; and
- compulsion not to have children, or compulsion to abort.[45]

Subsidiary issues also arise, among them the following:

- how to prevent or restrict further dissemination (for example, to insurers, police, researchers, governments or employers) of genetic information acquired by private reproductive clinics; and
- rights, if any, of children conceived as a result of a sperm or egg donation to learn the identity, or at least the genetic background, of the donor, and the potentially conflicting rights of the donor to have information about him or her kept confidential.

b. Employment

Employers may think they have a clear interest in genetic information about employees or job applicants. The information of interest might include risk factors for early onset Alzheimer's, heart disease, cancer, addiction, as well as some psychological traits and sensitivities to chemicals or other workplace contaminants. If the burden of health care costs

[45] The Privacy Commissioner of Canada, *Genetic Testing and Privacy,* above at 38.

shifts to the private sector, Canadian employers, like their American counterparts, may become much more interested in hiring only the healthiest employees with the "right" genetic stuff.

As of 1990, there appeared to be little, if any, genetic testing of employees or job applicants in Canada.[46] This is likely still true, in part because human rights legislation offers some protection against the genetic discrimination that might flow from testing. However, the American Civil Liberties Union (ACLU) has claimed that U.S. employers have substantially increased their use of genetic testing in employment in recent years, and that in 1997, 6 to 10 per cent of employers conducted genetic testing. The ACLU also reported "many documented cases" of genetic discrimination. It cited a survey of nearly 1,000 individuals who were at risk for genetic conditions. More than 22 per cent reported some form of discrimination based on their risk status.[47] The U.S. Department of Labor and several other US departments found genetic information to be a workplace issue that warranted federal legislative protection "to ensure that knowledge gained from genetic research is fully utilized to improve the health of Americans and not to discriminate against workers."[48]

At issue is the extent to which employers should be able to obtain and use personal genetic information to make decisions about employing individuals and assigning them to certain tasks. And to what extent are current human rights provisions against discrimination adequate to deal with workplace genetic testing?

c. Testing to Determine Eligibility for Services Such as Insurance and Credit

Genetic information, like other medical information indicating health status, can impede access to services such as insurance and credit. Equally, such information might make people more easily and less expensively insurable, and might also facilitate access to credit. As a result, genetic information may further separate those who have access to insurance, credit and other services from those who, because of their genetic makeup, do not.

[46] The Privacy Commissioner of Canada, *Genetic Testing and Privacy*, above, at 16.

[47] American Civil Liberties Association, "Genetic Discrimination in the Workplace Fact Sheet", ((2000) accessed October 23, 2000) [http://www.aclu.org/issues/worker/gdfactsheet.html (footnotes omitted)].

[48] Department of Labor, Department of Health and Human Services, Equal Employment Opportunity Commission, Department of Justice, *Genetic Information and the Workplace* (January 20, 1998).

Furthermore, the fear that genetic information being sought for health care reasons could be used to discriminate in insurance and in the provision of other services may discourage some people from seeking medically useful – in some cases, possibly life-saving – genetic testing.

Genetic information may have an impact on access to other services as well. For example, individuals with "superior" genetic traits might be singled out for special educational or vocational training, while those with "inferior" traits might see themselves denied or restricted in their access to such opportunities.

PART IV: RECOMMENDATIONS AND CONCLUSION

1. THE CENTRAL DILEMMA

At the heart of the debate over privacy, discrimination and genetic information is a concern that the failure to protect privacy and prevent discrimination will greatly diminish the potential for genetics to improve health and health care. Individuals may be afraid to undergo medically useful genetic testing or participate in socially useful genetic research for fear that this information may be used against them. Genetic inquiry may exacerbate loss of privacy and set the stage for greater discrimination. The most extreme uses of genetic information may go even beyond a deprivation of privacy and lead to genetically-based ethnic or racial "cleansing" or targetting of biological weapons.

Concerns about the misuse of genetic information are an extension of concerns about the misuse of personal information generally. Had society taken better care of non-genetic personal information to date, fears about the misuse of genetic information might be much less pronounced and much less justified.

2. ADDRESSING THE ISSUES

a. Education

Public education is essential to protect genetic privacy and prevent discrimination. Governments in particular have a duty to explain the uses of genetic information and their possible impact on society.

The 1992 report of the Privacy Commissioner of Canada, *Genetic Testing and Privacy*, is an example of the public education efforts that can help the public to grasp the significance of genetic privacy and discrimination issues. These in turn can lead to a more informed debate about the appropriate handling of genetic information. The work of the Canadian Genome Analysis and Technology Program and the 1999 *Final Report of the Advisory Council on Health Infostructure*, while not designed as vehicles for general public education, could also serve as useful starting points for the development of more easily understandable public education materials.

b. Control on Further Dissemination of Genetic Information

The key to benefitting from genetic information lies in controlling its uses beyond those relating to the health care of the individual to whom the information relates.

Particularly troubling, as seen with the US Department of Defense DNA collection program, is that DNA collected for one purpose (the identification of soldiers' remains) is also being made available for criminal investigations. Furthermore, in 1998, the FBI asked the U.S. government for extensive access to medical records without first getting patient permission, a request that would put numerous DNA databanks and patient records containing DNA profiles at the FBI's disposal.[49] A 1999 news report states that the Michigan Commission on Genetic Privacy has proposed that the state permanently keep DNA samples that had been taken for diagnosis of rare congenital diseases in newborns. The reason: the samples would prove valuable for law enforcement authorities and scientific research.[50] These developments will inevitably exert some influence on Canadian thinking about the handling of genetic information.

Calls for the extension of uses for genetic information that has been collected, and the seepage of practices and philosophies across our border, must be watched closely. It should not be the function of a DNA data bank assembled for health care or research to serve state authorities as a convenient object of plunder. Yet controls in Canada on such uses of DNA are weak.

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[49] American Civil Liberties Union, *The Year in Civil Liberties 1998.* Web site: http://www.aclu.org/library/ycl98.html (April 5, 1999).

[50] "Michigan Wants to Expand DNA Databank of All Newborns", The Detroit News, January 26, 1999.

Similarly, DNA collected and analyzed for health care purposes should not automatically be available for further uses, even research, if the DNA can be linked to an identifiable individual. The dissemination of this information to private commercial interests must also be tightly controlled.

Some legislation already protects genetic information. The *Charter of Rights* protects against abuses by government and against legislative measures that violate the guarantees of the *Charter*, but the full extent of that protection for genetic information will remain unclear for some time.[51] Legislation governing the private sector is inconsistent and incomplete. The recently enacted *Personal Information Protection and Electronic Documents Protection Act* will offer some protection against unwanted secondary uses of personal genetic information by commercial organizations, although the extent of those protections is not yet completely clear and will have to await judicial interpretation. As well, health information legislation such as that enacted recently in three provinces will help to promote "fair information practices" when dealing with genetic information. Statutory torts, codes of professional conduct, ethical guidelines and confidentiality provisions in health care legislation can also restrict the further uses of personal genetic information, although their effectiveness will vary.

Extensive legislative regulation and, in some cases, prohibitions, on secondary uses of personal information are indispensable for safeguarding the public interest and the privacy of individuals once information has been collected. The ultimate protection, however, may often lie in more carefully limiting the *initial* collection of personal genetic information.

c. Genetics-Specific or General Legislation?

Much of the discussion about protecting genetic information centres on whether specific genetic legislation is needed, or whether appropriately drafted general legislation will reduce discrimination and violations of genetic privacy.

[51] An explicit constitutional right of privacy, a right strongly supported by the Privacy Commissioner of Canada, would offer some comfort that DNA in the hands of government institutions would not without strong justification be made available for new uses.

Lemmens and Bahamin argue that regulating the use of genetic data, as a separate category of health-related information, could be impractical. "But it is important to point out the kinds of problems created. That might convince people of the need for stricter regulation of the use of medical information in general."[52]

Professor Mark Rothstein argues that "carefully crafted generic – rather than reflexively genetic – laws hold the greatest promise for protecting genetic secrets."[53] Rothstein also concludes that there is essentially no difference between ordinary medical information and genetic information, and that both types of information should be subject to the same protections.[54]

Indeed, rules governing personal health information generally can equally protect genetic information. Thus, legislation, policies and ethical guidelines for health information generally are relevant to genetic information. However, as stated earlier in this report, genetic information brings new intensity to the need to protect personal health information because of the abundance of sometimes highly sensitive personal information that genetic science produces or promises.

In some situations, legislative and other measures aimed at protecting health information generally may not deal appropriately with the peculiarities of genetic information. Among the issues that need to be addressed specifically because of the familial nature of genetic information are:

- whether and how to regulate the disclosure of genetic information about one person to that person's biological relatives where the information may be helpful to them; and
- whether it is possible and, if so, how to protect the right of biological relatives "not to know."

As well, legislative measures may be needed to protect the right "not to know" of minors, including measures to protect against newborns from being saddled with a genetics "report card" at birth.

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[52] Lemmens and Bahamin, above, at 150.

[53] Mark Rothstein, "Genetic Secrets: A Policy Framework," in Mark Rothstein, ed., *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (New Haven, Yale University Press, 1997) 451 at 459.

[54] Rothstein, above, at 458.

Forensic DNA legislation is another area where specific rules are needed. Canada has developed legislation dealing with the taking of DNA from criminal suspects and the establishment of DNA databanks relating to convicted offenders. However, this legislation must be carefully monitored to prevent an unwarranted enlargement of its scope.

d. Other Measures

Human reproduction: International and constitutional law offer some protection against discrimination by the state in matters relating to human reproduction. In particular, the right of privacy, preferably explicit, in conjunction with other rights, such as freedom of association, might forestall governments seeking access to genetic information for the sake of interfering with decisions relating to human reproduction. How well these legal measures will protect reproductive rights, particularly as governments are attempting to reduce health-care costs, must be watched. However, the greatest protection against state interference will come from preventing personal genetic information from falling into the hands of the state in the first place by enacting adequate restrictions on the state's collection of personal information.

Insurance: Genetic information highlights important concerns in insurance matters. As with other issues discussed in this report, it is not necessarily the genetic nature of the information, but rather the fact that the information can be used "against" the person, that demands attention. If individuals forego medically useful genetic testing because they fear losing their own or their family members' access to insurance, the main goal of genetic science – improved health care – is seriously undermined.

The solution – both to ensure access to the important good of insurance and to minimize privacy intrusions – may lie in prohibiting insurance companies from requiring medical information, genetic or non-genetic, for insurance policies of less than a stated amount. This would ensure that no Canadian could be denied basic insurance because of a genetic or non-genetic condition. Such rules would insulate insurance companies from the economic damage caused by adverse selection. It would eliminate the current disincentive for individuals to seek medically useful genetic testing.

For large amounts of insurance, it would continue to be appropriate for insurance companies to have access to relevant information about an applicant. However, companies should be prohibited from disclosing the information outside the insurance context. For example, they should be prohibited from disclosing genetic information to employers or other commercial interests.

Generally: The following measures would provide additional protection against the misuse of genetic information:

- ratifying the *Council of Europe and Convention on Human Rights and Biomedicine*.[55] As Professor Knoppers has argued, the debate about resolving genetics issues must be international. The solution may in part also need to be international;
- enacting statutory privacy torts in those provinces and territories that do not yet have them;
- encouraging provinces that have not yet done so to enact specific privacy and confidentiality protections for health care information;
- developing more detailed professional codes of conduct to deal with specific genetics issues, such as disclosure of information to family members; and
- enacting specific statutory protection in human rights codes against possible future disability.

[55] ETS No. 164 (1997).

3. CONCLUSION

Privacy and discrimination form only two sets of a complex array of issues surrounding genetics. Inattention to or deliberate neglect of privacy and discrimination issues can turn genetics from among the most promising advances in science into a powerful weapon for undermining fundamental human rights. The rapid advance of genetic science leaves little time to give real meaning to genetic privacy and to protect against widespread genetic discrimination.

POPULATION BIOBANKING IN CANADA: ETHICAL, LEGAL AND SOCIAL ISSUES

Document Prepared for the Canadian Biotechnology Advisory Committee

By

Lorraine Sheremeta

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TABLE OF CONTENTS

1. BACKGROUND

In 2002, the Canadian Biotechnology Advisory Committee commissioned a series of academic papers on issues associated with population genetic biobanks:

- Survey of National Approaches to the Development of Population Genetic Biobanks, by Mylène Deschênes and Geneviève Cardinal
- Toward a Comprehensive Information Privacy Regime for Research and Biobanks, by Michael Yeo[1]
- Whose Genes, Who's Safe, How Safe? Publics' and Professionals' Views of Biobanks, by Edna F. Einseidel

These papers, in conjunction with a presentation titled "Biobanks, Research and Privacy: Overview of Canadian Legislation" delivered by Patricia Kosseim at the Genome Canada GELS Winter Symposium held in Montreal, Quebec in February 2003, provide the basis from which this synthesis document is derived.[2] The purpose of this paper is to summarize and synthesize the salient ethical, legal and social issues that are relevant to biobanking in Canada. Where necessary, I have supplemented these works with other relevant material to expand and develop arguments as needed.

In addition to the materials noted above, there are at least four recent developments that warrant comment and inclusion in this synthesis document:

- The United Kingdom House of Commons Science and Technology Committee released a report titled "The Work of the Medical Research Council" [3] in which the committee levies harsh criticism at UK Biobank. In June 2003, the United Kingdom government publicly responded to the Committee's report. Appendix B contains a summary of the

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[1] YEO REVISED HIS WORK AFTER THE COMPLETION OF THIS PAPER AND IT IS NOW TITLED BIOBANK RESEARCH: THE CONFLICT BETWEEN PRIVACY AND ACCESS MADE EXPLICIT.

[2] The final versions of these four papers are available from the CBAC Web site: www.cbac-cccb.ca.

[3] United Kingdom, House of Commons Science and Technology Committee, "The Work of the Medical Research Council: Third Report of Session 2002-03," online: www.publications.parliament.uk/pa/cm200203/cmselect/cmsctech/132/132.pdf.

committee recommendations concerning Biobank along with the government responses[4] to those recommendations.

- In May 2003, in anticipation of bankruptcy, DNA Sciences Inc. sold substantially all of its assets, including its "Gene Trust" biobank, to Genaissance Pharmaceuticals.[5] The issues arising from this transaction are important and highlight the need for firms to employ defensive legal strategies to obviate the possibility that biobanks may be sold as assets in the case of financial hardship or bankruptcy proceedings. This situation raises general concerns over the sale or other disposition of biobanks and related data.
- In March 2003, the Canadian Biotechnology Secretariat released a new "wave" of public opinion research on biotechnology issues.[6] A portion of the survey data refers specifically to DNA mapping and patenting, and the data from this portion of the survey are included in the relevant sections of this paper.
- In August 2003, the Canadian Biotechnology Secretariat released a report titled "Public Opinion Research into Genetic Privacy Issues."[7] It is an important survey because it probes issues related to genetic privacy and biobanking and further contextualizes and reaffirms the tenor of the conclusions made by Professor Einsiedel concerning the publics' and professionals' views on biobanks.

In recent years, the storage and subsequent use of human biologic materials have become a hotly debated bioethical issue. Numerous professional organizations have issued statements that directly or indirectly touch on this issue.[8] At least three important developments

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[4] United Kingdom, Department of Trade and Industry, "Government Response to 'The Work of the Medical Research Council' Report by the House of Commons Science and Technology Select Committee (HC 132)," Cm 5834, June 2003.

[5] See DNA Sciences, "Genaissance Pharmaceuticals Enters into Agreement to Acquire Assets of DNA Sciences," News Release, April 1, 2003; online: www.dna.com/pressRelease/pressRelease.jsp?site=dna&link=20030331.htm; DNA Sciences, "Genaissance Pharmaceuticals' Acquisition of Substantially All of the Assets of DNA Sciences Is Approved," News Release, May 12, 2003, online: www.dna.com/pressRelease/pressRelease.jsp?site=dna&link=20030512.htm.

[6] Pollara and Earnscliffe Research, *Public Opinion Research into Biotechnology Issues in the United States and Canada* (Ottawa: Biotechnology Assistant Deputy Minister Coordinating Committee, 8th Wave, March 2003).

[7] Pollara and Earnscliffe Research and Communications, *Public Opinion Research into Genetic Privacy Issues* (Ottawa: Biotechnology Assistant Deputy Minister Coordinating Committee, March 2003).

[8] See, for example, Association of American Medical Colleges, "Policy Statement on Health Data Security, Patient Privacy, and the Use of Archival Patient Materials in Research" (1999), online: http://www.aamc.org/advocacy/issues/research/confplcv.htm; W. Grizzle et al., "Recommended Policies for Uses of Human Tissue in Research, Education and Quality Control" (1999), 123 Arch. Path. Lab. Med. 296; Medical Research Council, *Human Tissue and Biological Samples for Use in Research* (London: MRC, 1999), online: http://www.humgen.umontreal.ca; Working Party of the Royal College of Pathologists and the Institute of Biomedical Science, *The Retention and Storage of Pathological Records and Archives,* 2nd ed. (London: RCPATH,1999); Working Party of the Royal College of Pathologists and the Institute of Biomedical Science, *Consensus Statement of Recommended Policies for Uses of Human Tissue in Research, Education and Quality Control* (London: RCPATH,1999); World Health Organization, *Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services* (1999), WHO/HGN/GL/ETH/98.1, online: www.who.int/ncd/hgn/hgethic.htm; Human Genome Organization, "Statement on DNA Sampling: Control and Access" (1998), online: http://www.gene.ucl.ac.uk.hugo/sampling.html; Canadian Association of Pathologists, "Policy Statement and Guidelines for the Ethical Use of Human Tissue in Research" (1998), CAP Newsletter X; American Society of Human Genetics, "Statement on Informed Consent for Genetic Research" (1996), 59 Am. J. Hum. Genet. 471; Clayton et al.,

pertaining to the use of human tissue for clinical diagnosis and research have focussed attention on these issues. First, previously unforeseen uses have been identified for previously collected and stored tissue samples, and researchers are increasingly able to derive valuable genetic information from stored biological samples. Second, the research paradigm in genetics has shifted from linkage analysis of relatively small numbers of genetic samples to large-scale population genetic initiatives, which often involve the collection, storage and analysis of hundreds of thousands of samples. Third, with the advent of new, high throughput techniques to sequence DNA, there is a growing concern that the use of genetic and other medical information gleaned from these materials may be used in ways that violate individual privacy rights and could result in unfair discrimination against individuals and/or groups.

There is considerable optimism that society will benefit profoundly from innovations stemming from the Human Genome Project. It is hoped that analysis of data procured in large-scale population genetic studies will enable researchers to gain a better understanding of gene–environment interactions that are implicated in complex diseases such as heart disease, diabetes, Alzheimer's disease, multiple sclerosis and other commonly occurring human diseases. Numerous large-scale population genetic studies have been commenced and many more are at various stages of planning (see Table 1).

As with all technological advances, both risks and benefits are associated with population genetic studies. Despite the optimism that human health and well-being will ultimately be improved as a result of population genetic research, numerous ethical, legal and social concerns have been raised. For example, concerns regarding individual and group consent,[9] ownership of human biologic materials,[10] privacy and confidentiality,[11] genetic

"Informed Consent for Genetic Research on Stored Tissue Samples" (1995), 274 JAMA 1786; American College of Medical Geneticists, "Statement on Storage and use of Genetic Materials" (1995), 57 Am. J. Hum. Genet. 1499; American Society of Human Genetics, Ad Hoc Committee on DNA Technology, "DNA Banking and DNA Analysis: Points to Consider" (1988), 42 Am. J. Hum. Genet. 781.

[9] Laura M. Beskow et al., "Informed Consent for Population-Based Research Involving Genetics" (2001), 286 JAMA 2315; C. Weijer, "Benefit Sharing and Other Protections for Communities in Genetic Research" (2000), 58 Clin. Genet. 367; C. Weijer, G. Goldsand, E. J. Emanuel, "Protecting Communities in Research: Current Guidelines and Limits of Extrapolation" (1999), 23 Nat Genet 275; M. Deschênes, G. Cardinal, B. M. Knoppers et al., "Human Genetics Research DNA Banking and Consent: A Question of 'Form'?" (2001), 59 Clin. Genet. 221.

[10] Charlotte H. Harrison, "Neither Moore nor the Market: Alternative Models for Compensating Contributors of Human Tissue" (2002), Am. J. L. and Med. 77; E. Richard Gold, Body Parts: Property Rights and the Ownership of Human Biological Materials (Washington: Georgetown University Press, 1996); Moe Litman and Gerald Robertson, "The Common Law Status of Genetic Material," in edited by Bartha Maria Knoppers (Toronto: Emond Montgomery Publications, 1996).

[11] See Human Genetics Commission, Inside Information: Balancing Interests in the Use of Personal Genetic Data (London: Human Genetics Commission, May 2002); John A. Robertson, "Privacy Issues in Second Stage Genomics" (1999), 40 Jurimetrics 59; Lawrence O. Gostin and James G. Hodge, "Genetic Privacy and the Law: An End to Genetic Exceptionalism" (1999), 40 Jurimetrics 21; George J. Annas, "Privacy Rules for DNA Databanks: Protecting Coded 'Future Diaries'" (1993), 270 JAMA 2346.

discrimination and stigmatization[12] and eugenics[13] have been repeatedly raised. Academic researchers focussing on the ethical, legal and social issues of the Human Genome Project have initiated ample debate on these topics.

Table 1 –
Examples of Proposed and Existing Population Databases and Biobanks

SOME PROPOSED POPULATION DATABASES				
Project	**Company**	**DNA Sample Size**	**Budget**	**Status**
Icelandic Health Sector Database	deCODE Genetics	280 000	$212 million	Health database in 2003; 80 000 samples genotyped
Estonian Genome Project	eGeen Inc.	1 million	$150 million	3-year, $2.5-million pilot (10 000 donors) commenced Fall 2002
UK Biobank	not known	500 000	$66 million	Full enrolment in 2004
Marshfield Personalized Medicine		40 000	$3.8 million	Enrolling Fall 2003
National Children's Study (U.S.)		100 000	not known	Full study begins in 2004
Latvian Genome Database	not known	60 000	$1.7 million	Law passed in June; seeking funding
Quebec CARTaGENE	not known	50 000+	$19 million	Seeking funding
Existing Biobanks and/or Health Records				
Västerbotten, Sweden	UmanGenomics	80 000		Data use agreement with country in 2002
Mayo Clinic (U.S.)	not known	100 000		Prototype health database completed in July
European Prospective Investigation into Cancer and Nutrition (EPIC) (Europe)		350 000		Pooling data for cancer studies through consortium
Nurses' Health Study (U.S.)		63 000		
American Cancer Society Cancer Prevention Study (CPS-II) (U.S.)		110 000		
CDC National Health and Nutrition Examination Survey (NHANES III) (U.S.)		7 300		Proposals to use individual data requested fall 2002

Source: Jocelyn Kaiser, "Population Databases Boom, from Iceland to the U.S" (2002), 298 Science 1158–1161 at 1159.

[12] Henry T. Greely, "The Control of Genetic Research: Involving the 'Groups Between'" (1997), 33 Hous. L. Rev. 1397; Eric T. Juengst, "Group Identity and Human Diversity: Keeping Biology Straight from Culture" (1998), 63 Am. J. Hum. Genet. 673; H. Markel, "The Stigma of Disease: Implications of Genetic Screening" (1992), 93 Am. J. Med. 209.

[13] D. C. Wertz and J. C. Fletcher, "Ethical and Social Issues in Prenatal Sex Selection: A Survey of Geneticists in 37 Nations" (1998), 46 Soc. Sci. Med. 255; Anonymous, "Western Eyes on China's Eugenics Law" (1995), 346 Lancet 131; D. C. Wertz, "Did Eugenics Ever Die?" (2002), 3 Nat. Rev. Genet. 408.

At present, Canada is considering whether or not to commence a large-scale population genetic study.[14] The Canadian Lifelong Health Initiative remains at the planning stages, although it is planned to commence as early as 2005. The proposed study would follow the health of 30 000 infants from across Canada for a defined time period or for the lifetime of the infant. It has not gone unnoticed that Canada's national health system, like those in the United Kingdom, Estonia and Iceland, provides an ideal setting for a study of this type. The proposed study will involve early psychometric testing to evaluate intellectual, emotional and social development, as well as detailed environmental measures. The study is significantly smaller than many other large-scale initiatives. For example, UK Biobank expects to collect data on 500 000 individuals and the Estonian Genome Project hopes to collect data on approximately one million individuals (three-quarters of the country's 1.4 million population).

Because of the nature of the cohort, the planners of the proposed Canadian Lifelong Health Initiative will face numerous legal and ethical challenges. The fact that the research subjects are minors and will be unable to consent for many years is problematic and will require added precautions to ensure the strictest protection of the research subjects. There is general agreement that Canadian publics must be involved in meaningful consultation about the initiative and that the project planners must be responsive to concerns raised by the publics. Questions remain concerning how such consultation ought to proceed. The first issue that ought to be addressed through consultation is the determination of whether Canadians should promote population genetic research generally and, in particular, whether we should promote this particular initiative. The fact that the planners of the project deem the study meritorious does not necessarily mean that the general publics will be of the same opinion.

Failure to apply the highest scientific, legal and ethical standards to this initiative or similar programs will inevitably undermine public trust and confidence in scientific development and the products of such research.[15] The integrity of the project's development and of the research process itself is critical. Of the errors made in the development of the Icelandic health sector database, Wolfgang Edelstein aptly notes:

[14] Tom Spears, "Gene Study to Follow 30,000 Babies for Years," The Ottawa Citizen (February 7, 2003).

[15] William W. Lowrance, "The Promise of Human Genetic Databases: High Ethical as well as Scientific Standards are Needed" (2001), 322 BMJ 1009; D. Blumenthal, "Ethics Issues in Academic–Industry Relationships" (1996), 71 Acad. Med. 1291.

> *The procedural haste, the refusal to solicit the opinions of foreign experts (who have greater experience with industry/science/ethics conflicts), the unwillingness to take domestic criticism into account, the politicized and partisan debate in the case, the crude oversimplifications in the discussions and controversies over the biological processes basic to the inheritance of disease, the power of private interests, the plebiscitarian legitimation procedures in a case of subtle ethical, social and scientific controversy, all this is bound to raise a lot of misgivings, to say the least. It does not augur well for the search for consensual solutions of ethical conflicts in vulnerable domains of social life.*[16]

These issues also apply to other established projects. For example, the United Kingdom House of Commons Science and Technology Committee has recently censured the Medical Research Council in the United Kingdom for prematurely allocating funds to Biobank before questions over the project's value and methodology have been addressed.[17] In addition, the Committee alleges that "the scientific case for Biobank [was] put together by the funders to support a politically driven project."[18] The real debate in the United Kingdom over Biobank appears to have just begun.

If Canada wishes to pursue successful large-scale population genetic initiatives, it **must** heed these warnings.

"Biobank" Defined

In the previously commissioned works, and for purposes of this paper, a biobank is defined as a collection of physical specimens from which DNA can be derived, the data that have been derived from DNA samples, or both.

[16] Wolfgang Edelstein, "The Responsible Practice of Science: Remarks About the Cross Pressures of Scientific Progress and the Ethics of Research," December 1998, online: www.mannvernd.is/english/articles/we.twim.html.

[17] Supra, note 1 (Committee Report, Recommendation 31).

[18] Ibid.

Population genetic biobanks may be classified according to:

- their purpose (i.e., translational and basic science research, clinical research or clinical medicine)
- whether they already exist, or are prospectively planned
- whether they are temporary or permanent collections.

Population genetic research generally requires a biologic sample (frequently a blood sample or a buccal swab) from which the individual's genotype data are derived. Once these data are derived, they are stored in a database as "sequence data" and may also be linked to associated health information. In Canada, as in the United Kingdom, Iceland and Estonia, the linkage to health information would necessarily be facilitated by, and through, the publicly funded health care system.

The ethics of collection and storage of data and tissue in "biobanks" are frequently discussed in association with population genetic research. It should be recognized, however, that permanent or temporary collections of tissue are frequently established and maintained through of day-to-day clinical practice. For example human biologic materials are frequently obtained during diagnostic interventions or surgeries or where tissue or other material is obtained to determine the nature and extent of a disease. Where diseased tissue is removed after the diagnosis or treatment has been completed, a portion of the specimen is frequently retained — for future clinical, research, and/or legal purposes.[19] In addition, volunteers may donate their bodies, organs, blood or other biologic materials for educational purposes, transplantation or research.

The specimens procured may be stored in a variety of forms including slides, paraffin blocks, formalin-fixed specimens, tissue culture or extracted DNA. Biologic samples may be stored in clinical or research laboratories or in specialized tissue banks or DNA banks that operate to provide samples to public and private laboratories for both clinical and research purposes. While it is often important for routine clinical care that physicians have access to these stored samples, there are concerns about whether or not, and in what circumstances, existing specimens may be used for research purposes. The concern arises because the consent obtained at the time of the initial collection may not have specifically referred to future

[19] Purposes for which stored human biologic materials can be used include, but are not restricted to, the following: clinical use, development of research tools (e.g., cell lines, cloned genes, gene markers, purified proteins or isolated DNA) development of diagnostic or therapeutic commercial products quality control in health care delivery, and forensic identification.

research. The issues surrounding the collection, storage and use of human biological material obtained by researchers and by clinicians are frequently treated as discrete and separate, despite the inevitable overlap of clinical practice and research use.

It is important to note that prospectively created biobanks — like the one Canada is currently considering — will be less problematic than retrospective ones because participants can be appropriately informed about the uses to which their biological samples may be put; they can therefore consent to or refuse such uses. Having said this, there remains significant uncertainty about whether, or to what degree, patients can legally and ethically consent to unforeseen future uses of their biologic materials and associated data.[20] There is also uncertainty as to the legality of parental consent in the research setting. This paper focusses on the issues relevant to prospectively collected population genetic biobanks that will be used for translational and basic science research, as well as for clinical research. It is presumed that the collection would be of a permanent nature.

[20] Timothy Caulfield, Ross E. G. Upshur and Abdallah Daar, "DNA Databanks and Consent: A Suggested Policy Option Involving an Authorization Model" (2003), 4 BMC Medical Ethics, online: www.biomedcentral.com/1472-6939/4/1/; see also Henry T. Greely, "Breaking the Stalemate: A Prospective Regulatory Framework for Unforeseen Research Uses of Human Tissue Samples and Health Information" (1999), 34 Wake Forest L. Rev. 737.

2. OVERVIEW OF PREVIOUSLY COMMISSIONED WORKS

2.1 Survey of National Approaches to the Development of Population Genetic Biobanks, by Mylène Deschênes and Geneviève Cardinal

In this paper, Mylène Deschênes and Geneviève Cardinal consider various population genetic biobanks that have been developed around the world. Examples cited include CARTaGENE (Quebec), the Estonian Gene Bank, the Icelandic Health Sector Database (deCODE Genetics), Tonga (Autogen Ltd.), UK Biobank and the International Consortium's Genetic Variation Mapping Project (HAPMAP).[21] These projects represent a range of models that Canada may consider in planning population genetic research. For present purposes, this paper focusses on a smaller subset of the broader group of population studies examined by Deschênes and Cardinal, namely, the Icelandic Health Sector Database, the Estonian Genome Project and UK Biobank.

These particular initiatives are important because of the nature of the studies, the volume of information that exists about them, and what they reveal about the specific issues that must be addressed by Canadian policy makers. In this analysis, Deschênes and Cardinal identify nine major themes for Canada to consider with respect to biobanking. The substantive conclusions respecting each theme are summarized in the table below:

[21] For a recent comparison of several population genebanks, see, Melissa A. Austin, Sarah Harding Courtney McElroy, "Genebanks: A Comparison of Eight Proposed International Genetic Databases" (2003), 6 Community Genetics 37.

Table 2 –
Nine Major Themes for Canada to Consider in Developing a Biobanking Policy

THEME	KEY POINTS
Consultation (pp. 8–10)	▪ Fair and effective public consultation must occur prior to the initiation of any population genetic research.

Recruitment (pp. 10–12)	▪ Recruitment strategies are important and must seek to take into account the need to:
	  ▪ maintain individual autonomy, privacy and confidentiality in the recruitment process
	  ▪ ensure a fair distribution of risks and benefits among individuals in the subject population
	  ▪ ensure that the recruited subjects are representative of the overall population.
	▪ Recruitment strategies must be culturally sensitive and background information easy to understand. This means that multiple translations of information may be required.
	▪ In Canada, legitimate access to information held by a private or public institution could be obtained through mechanisms foreseen in privacy legislation; because of the variations in provincial legislation, a cautious review of jurisdictional requirements is needed.

Consent (pp. 12–17)	▪ Individual informed consent must be obtained to collect, to store and to use DNA samples and personal data in a population biobank. ▪ Consent forms should be adapted to reflect the benefits and risks for the population as well as the individual and should also address the issue of population benefit sharing. ▪ "Group consent" is neither practical nor desirable in the context of population genetic research. ▪ Public consultation should be undertaken and the opinion of the public should be properly considered if Canada intends to embark on a large-scale population genetic initiative.

Governance (pp. 17–22)	▪ At present, there exists no coherent legal and ethical framework to accommodate the peculiarities of biobanking.
	▪ Biobanks, though traditionally conceived of as pure research and therefore public sector endeavour, are becoming increasingly mixed with private industry.
	▪ The commercial aspects of biobanking must be carefully managed so as not to undermine public support for such endeavours.
	▪ A governance scheme must be transparent and accountable and must inspire trust in all stakeholders.
	▪ There is a need to create an independent organization that would be responsible for overseeing the overall project and surveillance activities. Surveillance must focus on both management and operational issues.
	▪ Ethical approval and monitoring of population genetic research should be required, regardless of who performs the research or where the research is performed.
	▪ The principles contained in the Tri-Council Policy Statement (TCPS)[22] should be applicable equally to public and private entities.
	▪ However, specific concerns have been identified with regard to the TCPS:
	  ▪ it does not apply to privately funded research
	  ▪ the point at which a research ethics board should be consulted is not clear, nor is the proper composition of such a board
	  ▪ the longitudinal nature of population genetic research projects requires the development and implementation of long-term monitoring and oversight mechanisms, an issue not adequately addressed by the TCPS
	  ▪ the added difficulties posed by multi-centre trials with respect to ethical approval and oversight must be taken into account.

[22] Medical Research Council, Natural Sciences and Engineering Research Council of Canada and the Social Sciences and Humanities Research Council of Canada, Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans (Ottawa: Public Works and Government Services, 1998).

THEME	KEY POINTS
Commercialization (pp. 23–26)	▪ Potential commercial application: populations and individual research subjects must be made aware, during the informed consent process, that research may lead to the development of commercial products, the filing of patent applications and the generation of revenue. ▪ Benefit sharing: the concept of benefit sharing stems from the notion that the human genome is a collective and vital interest of humankind and that the benefits and burdens of exploiting and sustaining the resource ought to be universally shared. Benefits should not be construed narrowly. The HUGO Ethics Committee Statement on Benefit Sharing states that they may include "agreements with individuals, families, groups, communities or populations that foresee technology transfer, local training, joint ventures, provision of health care or information infrastructures, reimbursement of costs, or the possible use of a percentage of any royalties for humanitarian purposes."[23] ▪ Freedom of access: liberal though carefully controlled access by researchers to population genetic data is in the best interests of subject populations and accords with the traditional norms of science. Exclusive licensing of biobanks to a single or to a select few commercial entities is ethically and legally problematic. ▪ Conflict of interest: the interests of the subject population must be appropriately represented in commercial transactions that involve data derived from population genetic research.

Privacy (pp. 27–30)	▪ Because of the highly personal and sensitive nature of the information stored in biobanks and associated databases and because of the volume of the data to be amassed, very strict physical, procedural and electronic safeguards are required to protect the information that is entrusted by participants to researchers:
	  ▪ biobanks must be physically protected; the research protocol should outline the exact storage conditions, including security measures and access requirements
	  ▪ access to biobanks for non-medical research purposes (i.e., to law enforcement agencies) should be prohibited or participants must be made aware of those circumstances in which data may be released to third parties
	  ▪ access to biobank data for research purposes should be controlled by a guardian or custodian that does not itself perform research and that will ensure the required level of confidentiality is met prior to release of data
	  ▪ precautionary measures must be developed for data processing and linkage between databases
	  ▪ staff must be made aware of their **duty** to protect the privacy and confidentiality of biobank data and that failure to carry out that duty will result in appropriate disciplinary action.
	▪ An independent authority (i.e., the Privacy Commissioner or other mandated authority) should play a role in supervising data protection and ensuring compliance with privacy laws and regulations.
	▪ Criminal or other sanctions may be necessary and appropriate in circumstances of wrongful disclosure of confidential data. Estonia and Iceland provide harsh penalties including fines and/or imprisonment for wrongful disclosure of personal biobank-related information. In Estonia, participants whose data have been wrongfully disclosed may request the complete destruction of their biological samples and related information**.**

Communication of research results (pp. 31–34)	▪ Populations involved in large-scale genetic research should be regularly informed of research results. Frequent communication to the study participants is a way to show respect and appreciation for their efforts and will foster mutual trust between researchers and participants. ▪ Results of population genetic research must be communicated in a way that will encourage understanding of the information; researchers must avoid the adverse effects of public disclosure of research results without full explanations. ▪ The scientific community is obliged to participate in a dialogue concerning the results of research – to ensure that the results are scientifically accurate and are understood by the populations, families and individuals that participate in and are affected by the research.

[23] HUGO Ethics Committee. "HUGO Statement on Benefit-Sharing" (2000), 6 Genome Digest 7-9.

THEME	KEY POINTS

Welfare of the population (pp. 34–37)	▪ Thorough reflection must be undertaken on the relative risks and benefits of population genetic research.
	▪ Because population genetic research poses risks to an entire population, it is important that societal benefits outweigh societal risks:
	  ▪ potential benefits of population genetic research include health benefits, improved population health care strategies, the creation of a repository of information and biological samples that may be useful for future research, economic benefits and collaborative research ventures
	  ▪ potential risks of population genetic research include adverse economic effects, discrimination (employment, insurance), inappropriate disclosure and/or use of sensitive and personal information and the possibility that human genetic material could inappropriately be used for human reproductive cloning or bioterrorism.

Welfare of Humanity (pp. 37–38)	▪ The universality of the human genome and the bioethical principles of beneficence and justice demand the sharing of knowledge about the human genome. ▪ Scientific discoveries that depend on the human genome should be channelled toward the improvement of global health; benefits should not accrue preferentially to the developed world at the expense of the developing world. ▪ Benefit-sharing mechanisms, the granting of unrestricted access to genetic sequence data by researchers, and mechanisms to limit negative effects of conflicts of interest will help to ensure that the benefits of human population genetic research will accrue fairly to participating populations and to the global community.

2.2 Toward a Comprehensive Information Privacy Regime for Research and Biobanks, by Michael Yeo

In his paper, Professor Michael Yeo paints a view of society in which individual privacy is increasingly "under siege from a variety of interests which may or may not be noble and benign to greater and lesser degrees" (p. 5). Biobanks are but one of the many ways that personal information is collected, stored and used (whether appropriately or inappropriately).

The fact that population genetic data are typically stored as a searchable and permanent record means that there is some degree of risk that the information contained in biobanks could be used in ways that are contrary to the interests of the individuals volunteering for the research and to whom the data refer, or to the larger groups to which the individuals belong.

Because the issues raised by population genetic research and biobanks are societal in nature, they demand broad community discussion to ensure that societal benefit outweighs societal risk. These discussions must be resolved prior to the initiation of a population-based biobank and must serve to ensure that there is a measurable balance struck that will ensure transparency and accountability (p. 3). Though many of the putative risks associated with biobanks are indirect and speculative, they are probably real and must be taken seriously, and

the broadest concerns of the public must be considered. That said, the growing corpus of provincial privacy legislation dealing specifically with health information attests to the fact that privacy is an issue of significant concern in Canadian society. Cutbacks to government funding of health care, in combination with increased demands for access to data, are creating additional challenges to the system. Despite this, the need for highly sophisticated and secure linkages between biobanks and publicly collected health information must not be underestimated.

Yeo places health professionals and medical researchers in the general category of the many who seek access to information. He notes that they acknowledge the value of privacy protection of personal health information, yet argue that their specific use of protected information should be treated as an exception to the general rule. They accept that privacy is important, but justify their proposed use as being "very important" and clearly "in the public interest" (p. 5, citing Freeman and Robbins). Some expect their uses to be exempted from the consent requirement; most expect that any requirements imposed will not unduly impede access to the information they seek. Medical researchers are unlikely to view themselves in the broader context of the many others with competing (and virtually identical) arguments to obtain access to information.

With respect to the various interpretations of privacy, Yeo notes that there is no single definition that captures all that the concept of privacy encompasses. He describes privacy as an "essentially contested concept," providing a number of examples (at p. 21). Definitions of privacy provided by Yeo include:

- "The right to be let alone." (Warren and Brandeis, 1890)
- "The claim of individuals, groups or institutions to determine for themselves when, how, and to what extent information about them is communicated to others." (Alan Westin, 1984)
- "The extent to which we are known to others, the extent to which others have physical access to us, and the extent to which we are the subject of others' attention." (Gavison, 1984)
- "The control we have over information about ourselves." (Fried, 1984)

Yeo notes that there are numerous concepts related to and overlapping with the concept of privacy, including ownership, autonomy, dignity, trespass, intrusion, intimacy, anonymity, secrecy, security, solitude and inviolate personality (p. 21). It can be argued that genetic information encompasses many of these; however, it can also be argued that these data transcend this list because they are related not only to the individual from whom they were obtained, but also to his or her family, as well as to the broader community or communities to which the individual belongs. Significantly, Yeo notes:

> *Because the concept of privacy is as rich, evocative and amorphous as it is, it serves as a kind of magnet for anxieties and concerns about rapid social and technological change that are fuzzy and hard to articulate. Its evocativeness and amorphousness is no doubt a detriment in some respects, but can also be seen as a virtue to the extent that it enables voice for worries that otherwise might not find a name or expression. (p. 23)*

Because the sense of self varies considerably from culture to culture, differences between groups must also be taken into account when considering the impact of population genetic research. For example, Dr. Frank Dukepoo, an aboriginal geneticist, has stated:

> *To us, any part of ourselves is sacred. Scientists say it's just DNA. For an Indian, it is not just DNA, it's part of a person, it is sacred, with deep religious significance. It is part of the essence of a person. (Yeo, p. 10)*

In recognition of the fundamental tension that exists between privacy and research, Yeo examines emerging developments in research and biobanking from the perspective of privacy. He attempts to create a framework within which one can "identify, discuss and debate privacy-related issues and concerns." What emerges is a recognition that privacy has several different meanings and encompasses a variety of values. Because of this, individuals will have differing views as to the proper balance between strict privacy and the free sharing of genetic data and health information to facilitate medical research. Yeo elaborates on two main perspectives on privacy: self-determination and benign stewardship. He suggests that both perspectives must be taken into account in the development of an accountable regulatory framework that will protect privacy. Table 3 provides a summary of these two perspectives.

Table 3 –
Summary of the Principles and Ideologies to Consider in Developing a Comprehensive Regulatory Framework for Biobanking in Canada

<table>
<tr><th colspan="2">FAIR INFORMATION PRINCIPLES</th></tr>
<tr><td>Accountability
Identifying Purposes
Consent
Limiting Collection
Limiting Use, Disclosure and Retention</td><td>Accuracy
Safeguards
Openness
Individual Access
Challenging Compliance</td></tr>
<tr><td>Self-determination
Liberalism
Deontology
Autonomy
Rights
Consent-based
Accountability</td><td>Benign Stewardship
Communitarianism
Utilitarianism
Beneficence
Non-maleficence
Utility
Safeguards</td></tr>
<tr><td>Self-determination Frame
The individual has a right to control the collection, use, and access to his or her personal information.
The individual also has a right to know:
who else may obtain access
circumstances in which information may be collected, used, accessed or disclosed without authorization
potential harms that may accrue if information is disclosed
the practices and policies of the data steward
safeguards that are in place to address the risks.
Individuals have the right to participate as citizens in a free and democratic society and not merely as “data subjects.”
Accountability</td><td>Safeguarding Frame
Protocols in place to authorize information use and mechanisms to ensure that information flows as authorized
Confidentiality agreements, oaths and promises elicited from those with access to information; penalties against unauthorized use or access
Training of information users and other persons who are part of the regime with respect to protocols, roles and responsibilities
Security measures, including locks, passwords, encryption, firewalls, etc. to prevent unauthorized users from accessing information
Incorporation of privacy enhancing technologies such as audit trails
Procedures for anonymizing, deidentifying or coding information
Internal policy standards that are appropriately communicated and publicized
External regulatory standards (law, policy, professional codes) to which the steward can be held accountable
Institutional privacy officers or committees to monitor compliance with organizational policy and external regulatory standards
Independent, external oversight bodies (e.g., privacy commissioner)
Proxy bodies (e.g., ethics committees or community panels) standing in the place of individuals and communities whose information is being held in trust.
Privacy impact assessments to map data flow and to assess safeguards and policies to enable the steward to perfect the information regime and to promote transparency
Accountability</td></tr>
<tr><th colspan="2">COMPREHENSIVE REGULATORY FRAMEWORK</th></tr>
</table>

A comprehensive regulatory framework for privacy will, of necessity, include provisions addressing the two main sorts of issues. For example, while a volunteer may have the right to withdraw from participation and may request to have his or her biologic sample and associated data destroyed, there are corollary safeguarding provisions that must be in place to make withdrawal possible. Yeo creates a compelling argument that any privacy policy that attempts to define the rights and responsibilities associated with genetic information and biobanks must seek to integrate the principles enshrined in both perspectives.

The risks associated with biobank research and the responsibilities incumbent on the researcher may be abstracted from Yeo's paper and are summarized in Table 4.

Table 4 –
Risks and Responsibilities Associated with Population Genetic Research

RISK	RESPONSIBILITY
▪ Adverse societal effects ▪ Loss of individual privacy ▪ Potential breadth of research questions ▪ Scale of project	▪ Need for public consultation ▪ Need for: transparency, accountability and reassessment of the current governance framework ▪ Need to consider impact on informed consent norms ▪ Need to explore public funding and innovation strategies

In summary, Professor Yeo reaches the following conclusions:

- Large-scale biobanks are more akin to business organizations than to research structures as traditionally conceived, and the existing legal and ethical framework that governs research is not adequate.
- Biobanking and applied genetics research differ significantly from other research endeavours that involve human subjects. These differences warrant amendments to the current legal and ethical regime or require the development of a separate regulatory regime. Specifically, in Canada, consent issues are not adequately addressed in the existing regulatory framework.
- Biobanking requires the development of an accountable regulatory framework that incorporates the legal and ethical norms governing human subject research and the evolving ethical norms of corporate governance.

2.3 Whose Genes, Who's Safe, How Safe? Publics' and Professionals' Views of Biobanks, by Edna Einsiedel

Professor Edna Einsiedel's paper seeks to identify the views of the publics and of professionals that are relevant to genetic technologies generally, and to biobanks in particular. The data presented in this paper are derived largely from papers published in peer-reviewed journals (predominantly *Science* and *Nature*) or from published reports that have emerged as a result of the development of the Icelandic Health Sector Database and UK Biobank. Einsiedel's paper is composed of two distinct parts. The first focusses on the views of the general public and advocacy organizations; these groups comprise the "unorganized public." The second part of the paper concerns the views of professionals, who are defined to include geneticists, genetic researchers, genetic counselors, research coordinators, members of research ethics boards or institutional review boards, physicians, ethicists, legal experts and epidemiologists. Table 5 lists specific issues that are considered by Einsiedel with respect to public and professional perspectives.

Table 5 –
Public and Professional Perspectives Relating to Biobanks: Summary of Issues Considered

PERSPECTIVE OF THE PUBLICS	PERSPECTIVES OF PROFESSIONALS

▪ Public awareness and understanding
▪ The need for more information
▪ The role of the media
▪ Recruitment and participation
▪ Informed consent
▪ Feedback
▪ Confidentiality
▪ Ownership and control of databases
▪ Commercialization of genetic information
▪ Human rights issues
	▪ Informed consent
	▪ Consent to future uses
	▪ Confidentiality and privacy
	▪ Feedback
	▪ Human rights issues
	▪ Individual rights versus community rights
	▪ Commodification of the human body
	▪ Questions of identity
	▪ Reductionism
	▪ Concerns of professionals:
	  ▪ commercialization
	  ▪ non-research use of databases
	  ▪ governance issues

Specific data have been extracted from Einsiedel's paper and incorporated into topical summary tables in the relevant subsections of Section 4 of this paper.

While providing a comprehensive collection of snapshots of publics' and professionals' views on specific issues from the international perspective that are useful to consider, Professor Einsiedel notes that it is critical that Canada endeavour to gain an understanding of the issues from the Canadian perspective. Canada is a large, geographically diverse territory with a diverse population, but within this larger heterogeneous community there are small homogenous subpopulations (i.e., French Canadians, Newfoundlanders and various aboriginal communities). Disparities in local needs and interests must be well understood if Canada is to embark on successful large-scale population genetic initiatives.
In summary, Professor Einsiedel concludes that:

- If publics are to be meaningfully engaged in debates about genetic technologies, they need to be able to:
    - assess benefits and risks of genetic technologies in a rational way
    - understand the limits of the science
    - consider the ethical, legal and social dimensions of these issues.
- Public education initiatives must provide genuine opportunities for engagement and deliberation and permit individuals to reach an informed opinion about the issues.
- "Meaningful dialogue" (i.e., that which is "mutually informative, thoughtful, honest, and carries the possibility of being mutually transformative") should be fostered between scientists, other stakeholders and the public (p. 41, citing McLean, 2001).
- Canada may wish to consider developing a communication plan similar to that recommended by the Centers for Disease Control in the U.S. to "assess 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" (p. 40).
- Public engagement is a critical requirement **before** Canada embarks on a large-scale population genetic research initiative: meaningful public involvement demands posing the question of whether such a venture is worthwhile and is an appropriate use of public funds (p. 42).

- For individuals to make an informed decision about whether to participate in a large-scale population genetics initiative, they must have information regarding the purpose(s) of the research, the conditions for maintaining privacy and confidentiality, consent conditions for future access and/or secondary uses of the data, conditions for storage and maintenance, oversight mechanisms, and the potential for commercialization (p. 42).
- Because the implications of population genetics research are far-reaching, it is important that if Canada opts to develop a large-scale initiative (or initiatives), the governance framework should include an independent oversight committee with public representation (p. 42).
- There may be a role for a national standing oversight panel (research ethics board) to review and monitor biobank research, keep track of the project history, develop and maintain periodic reports, serve as a resource for ethical, legal and social issues and provide a forum for ongoing national discussion (p. 44, citing Martin, 2001).

3. NEW DEVELOPMENTS

3.1 United Kingdom House of Commons Science and Technology Committee Report

On March 25, 2003, the United Kingdom House of Commons Science and Technology Committee released its report titled "The Work of the Medical Research Council."[24] Although the report is based on an evaluation of the scope of work of the Medical Research Council (MRC), it is devoted in large part to the Biobank project. The report alleges that, within the MRC, there is generally evidence of poor financial management, poor planning and too many funds being committed over too long periods, leading to large numbers of top-quality grant proposals being turned down. In this report, the MRC is accused of implementing misguided strategies for its research support that have resulted in discrimination against young researchers and some disciplines.

The committee specifically alleges that UK Biobank is a "politically driven project"[25] that was established without full confidence of the research community and the public. In fact, the committee alleges that funds (including a 45-million-pound grant from the MRC were allocated to the project before the scientific questions over its value and methodology were fully addressed[26] and that consultation for Biobank was "a bolt-on activity to secure widespread support rather than a genuine attempt to build a consensus on the project's aims and methods."[27] UK Biobank is described as a top-down initiative, the merits of which have not been properly balanced against other potential funding options.

The United Kingdom government responded to the committee's criticisms in June 2003.[28] In essence, while recognizing the challenges facing the MRC and the general need to improve in the areas of financial management, long-term planning, evaluation and communications, the government defended the MRC's research strategies on the basis that they were developed by the MRC in consultation with a broad range of organizations.

[24] Supra, note 1 (Committee Report).

[25] Ibid. para 58.

[26] Ibid. para 57.

[27] Ibid. para 65.

[28] Supra, note 2 (Government Response).

Specific criticisms levied by the committee with respect to UK Biobank and the government responses to those criticisms are appended to this paper (see Appendix B). In addition, specific issues are highlighted in the relevant subsections of Section 4 of this paper.

3.2 DNA Sciences Inc. Sale of the Gene Trust to Genaissance Pharmaceuticals

In recent years, the United States has seen an emergence in the development of commercial biobanks that operate to meet the need growing need for human tissue, DNA and associated data in the public and private sectors.[29] Detractors are concerned about the ethical appropriateness of the free-market approach in this field[30] and about the lack of protection for human research participants in the United States.[31] Gaps in the federal regulatory system effectively make federal research guidelines inapplicable to privately funded research or commercial endeavours.[32]

Whether private or public entities are involved in commercial biobanking, there are real opportunities for approaches that are antithetical to modern ethical standards. One news report describes the sale of neonatal blood samples by the South Carolina state government to a private company for use in the development of genetic testing kits and also to the law enforcement division for baseline studies of DNA markers.[33]

Without the use of specific legal protections, commercial biobanks may be unable to adequately protect the interests of biobank participants. The sale of DNA Sciences Inc. reveals that this may be especially true in times of financial hardship or when bankruptcy proceedings are commenced.

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[29] Eliot Marshall, "Company Plans to Bank Human DNA Profiles" (2001), 291 Science 575. This article focuses on the business strategy of First Genetic Trust Inc. It sees itself as "an intermediary between patients and researchers." Individuals permit the company to store their genetic information in a confidential database for use in clinical research; visit their corporate Web site at: www.firstgenetic.net/. Another company, Ardais Inc. has a similar business strategy; visit their corporate Web site at: at www.ardais.com.

[30] Mary R. Anderlik, "Commercial Biobanks and Genetic Research: Banking Without Checks?" presentation delivered at the 3rd International DNA Sampling Conference, September 5–8, 2002, Montreal, Quebec.

[31] Eric M. Meslin, "Raising the Bar in Research Ethics: Traditional Obligations are Not Enough" (2002), 112 Postgraduate Medicine 5.

[32] Ibid. See also "Balancing Privacy and Biotechnology," Business Week, editorial (15 April 2002).

[33] Dana Hawkins, "Keeping Secrets," U.S. News and World Report (12 February 2002). See also Michael J. Trebilcock and Edward M. Iacobucci, "Privatization and Accountability" (2003) Harvard L. Rev. 1422. The authors note that people too often incorrectly conclude that flaws in the private market imply the need to maintain public sector influence. They propose that, in discussing the merits of public and private action, the analysis must be relative. They also suggest that private market shortcomings "pale in comparison to the flaws associated with public provision or public oversight of private actors."

DNA Sciences, Inc.,[34] now defunct, was an applied genetics company based in California that focussed its business development on the discovery and commercialization of DNA-based diagnostic tests. By creating the Gene Trust,[35] DNA Sciences Inc. sought to establish a database of information about individuals that included physical characteristics, health histories and ongoing data concerning medical treatment and effectiveness. To facilitate its work, DNA Sciences utilized the Internet to attract Gene Trust volunteers who were asked to provide contact information and a personal family health history. If found to be an appropriate volunteer, informed consent was elicited and a blood sample was obtained and analyzed.[36] The DNA Sciences Web site reported that more than 10 000 participants from all 50 states were registered in the Gene Trust.[37]

As part of the recruitment strategy, the "Gene Trust Bill of Rights"[38] assured participants that personally identifying genetic information would never be sold or shared with anyone outside the Gene Trust; once collected, information was to be made anonymous and the Gene Trust researchers would use anonymous data only. Genetic information would never be supplied to employers or insurance companies, human cloning would not be undertaken and DNA Sciences would not be associated with any such practices. Participants were free to withdraw at any time, for any reason and without penalty. [39]

DNA Sciences assured participants that personally identifying information would never be shared "with any person or entity outside DNA Sciences without your express consent, unless legally required to do so." [40] The privacy statement is laudable but likely irrelevant in light of the fact that the consent form signed by participants contained a provision permitting DNA Sciences to transfer samples and anonymized medical data to a third party.[41] On May 15, 2003, Genaissance Pharmaceuticals announced that it had entered into an agreement to

[34] Supra, note 3 (DNA Sciences) online: DNA Sciences 65.161.124.110/home/home.jsp?site=dna&link=Home.htm. DNA Sciences Inc., was incorporated in 1998. Its corporate office is located in Fremont, California.

[35] Ibid., online: DNA Sciences 65.161.124.110/sectionHome/sectionHome.jsp?site=dna&link=TheDNASciencesGeneTrustProject.htm.

[36] Ibid., online: DNA Sciences 65.161.124.110/sectionHome/sectionHome.jsp?site=dna&link=Origins.htm.

[37] Ibid., online: DNA Sciences www.dna.com/sectionHome/sectionHome.jsp?site=dna&ink=Howweredoing.htm.

[38] Ibid., "The Gene Trust Bill of Rights," online: DNA Sciences, http://www.dna.com/sectionHome/SectionHome.jsp?site=dna&link=TheGeneTrustBillofRights.htm.

[39] Ibid.

[40] Ibid., "DNA Sciences Privacy Policy," Version: 4, Updated: 11-5-02, online: DNA Sciences http://www.dna.com/privacy/Page/privacyPage.jsp?site=dna&link=PrivacyStatement.htm.

[41] Personal communication, Melodie Henderson, Vice President, Intellectual Capital and Licensing, Genaissance Pharmaceuticals, Inc.

acquire substantially all of the assets of DNA Sciences.[42] This agreement includes the Gene Trust DNA samples, the anonymized medical history data, and the computer systems that hold personally identifying data on the Gene Trust donors.[43] At this time, Genaissance Pharmaceuticals does not intend to continue the Gene Trust. The situation, however, raises many legal and ethical questions that must be considered by Canadian policy makers.

In Canada, corporate entities could be used as vehicles to "own" public biobanks. The possibility that the corporation could go bankrupt or would be wound up must be considered at the outset to ensure that human biological samples, DNA sequence data, medical history data and other personal information will not be inappropriately "sold off" to a third party. As part of this framework, a legal trust could be employed to protect a biobank from falling into a corporation's general asset pool and sold off. The biobank could, in essence, be held by the corporation in trust for (i.e., for benefit of) the participants whose data are held in the biobank. In Canada, issues of corporate governance need to be further explored in relation to biobanks.

3.3 Public Opinion Research into Biotechnology Issues – Eighth Wave

Since 1999, the Government of Canada's Canadian Biotechnology Secretariat and its partners have maintained a large-scale program of public opinion research involving 10 public opinion surveys and more than 75 groups. These studies represent North America's largest and most comprehensive investigation into attitudes about biotechnology and the public policy that surrounds it. The program is designed to produce two waves of research each year, with a large tracking component and chapters of more intensive inquiry into specific issues like "genetic privacy." Results have been remarkably consistent since the inception of the research program.

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[42] Supra, note 3. See, DNA Sciences, "Genaissance Pharmaceuticals Enters into Agreement to Acquire Assets of DNA Sciences," News Release, April 1, 2003, online: DNA Sciences www.dna.com/pressRelease/pressRelease.jsp?site=dna&link=20030331.htm; DNA Sciences, Genaissance Pharmaceuticals' Acquisition of Substantially All of the Assets of DNA Sciences is Approved, May 12, 2003, online: DNA Sciences www.dna.com/pressRelease/pressRelease.jsp?site=dna&link=20030512.htm.

[43] Supra, note 39.

The eighth wave of research, completed in March 2003, represents a cross-national study of attitudes toward biotechnology in Canada and the United States. Released in August 2003, the research is based on telephone surveys of 1000 Americans and 600 Canadians using a single questionnaire. The survey was designed to meet two main objectives:

- track public sentiment on a range of biotech issues in the Canada, using a baseline of data developed in previous waves of research
- compare attitudes among Americans and Canadians.

This survey is of interest because it specifically asks several questions with respect to DNA mapping and the patenting of genes. Specifically, Canadians are described as "cautiously supportive of progress and of science." Seventy-eight percent of Canadians see more benefits than drawbacks in the areas of DNA mapping. Approximately half are uncomfortable with the idea of patenting in the area of biotechnology. Results of this survey are incorporated into the relevant subsections of Section 4 of this paper.

3.4 Public Opinion Research into Genetic Privacy Issues

POLLARA Inc. and Earnscliffe Research and Communications jointly performed surveys and hosted two focus groups to elicit public opinion on privacy issues associated with genetic information in February and March 2003. The results of this research were released in August 2003. The study is important because it helps to put into context the findings presented by Edna Einsiedel, and it deals specifically with issues surrounding biobanks.

The survey reveals, not surprisingly, that "[t]he concept of biobanks is not yet fully formed in the [Canadian] public mind" (p. 10). In summary, the survey reached the following conclusions:

- People do not understand how population health or genetic studies are conducted.
- People are increasingly aware of research strategies to trace genetic histories through families and to gather data from related people.

- People think of biobanks in terms of information and not in terms of physical samples.
- People have difficulty contemplating the collection and storage of large volumes of genetic information.
- People have no idea whether biobanks are prevalent or who might be administering them.
- People presume the regulations governing biobanks would tend to be relatively lax, largely because biobanking is a new phenomenon. (pp. 10–11)

Results of this survey are incorporated into the relevant subsections of Section 4 of this paper.

4. DISCUSSION: MAJOR ISSUES FOR CANADA TO CONSIDER RE BIOBANKING POLICY

This discussion provides a synthesis of the major ideas gleaned from the previously commissioned works. The subsections on privacy and confidentiality depend heavily upon multiple external sources. The following issues are addressed here:

- consultation, education and the role of the media
- recruitment
- privacy and confidentiality
- informed consent and communication of research results
- commercialization
- governance issues.

4.1 Consultation, Education and the Role of the Media

Authors of the previously commissioned papers have unanimously concluded that prior public consultation is a necessary first step if Canada is to be committed to the idea of initiating a large-scale national biobank project (Deschênes and Cardinal, p. 10; Einseidel, p. 44–45; Yeo, p. 58). Successful population genetic research depends directly on public goodwill and trust. The processes of implementation must therefore be based on transparency, public discussion and genuine debate (Deschênes and Cardinal, p. 8). Consultation is important for a number of reasons, not least of which is that it may help to uncover specific weaknesses inherent in the proposed study design, particularly those elements that, if not corrected, will undermine effective communication between researchers and the subject population and the population more broadly. In addition, although not part of the informed consent process, consultation will enable thoughtful reflection by potential research participants and the broader community on relevant issues and will facilitate a more robust informed consent than would otherwise be possible. Consultation has the potential to open a meaningful dialogue between the research participants and the research team and to facilitate meaningful democratic participation of citizens.

Einsiedel notes that the development of the Icelandic health sector database set off an international debate over issues relating to genetic information and biobanks (p. 7) and that, largely as a result of the Icelandic controversy, UK Biobank was conceived by the MRC and the Wellcome Trust with the specific intent of proactively understanding and responding to stakeholder concerns. Accordingly, a number of consultation studies were commissioned and reports published.[44]

Despite these consultative efforts, a recent report of the United Kingdom House of Commons Science and Technology Committee[45] harshly criticizes the consultation process employed by the MRC (see also Yeo, pp. 57–58**)**. In light of this criticism, careful consideration must be given to the development of consultative strategies that will yield information that will impact project planning, that will engender a collaborative relationship between researchers and the population and will not be viewed as a means to further a political agenda (Deschênes and Cardinal, p. 10). If executed in bad faith or if poorly performed, consultation will inevitably undermine trust between the numerous publics, the scientific community and policy makers.

To be effective, consultation must be broadly reflective of stakeholder concerns. A non-exhaustive list of stakeholders in the context of biobanking includes: the participants (and their relatives or fellow "group" members), the researchers and the research community, health care providers, agencies that fund research, regulating bodies, other users of data, special interest groups and the media.[46] Yeo concurs with the recommendation of Quebec's Commission de l'ethique de la science et de la technologie that "all population genetic databases for mapping a population's genes or conducting research on population genetics first be submitted to an informed public to actively involve them in the decision-making process" (Yeo, p. 56). Table 6 provides a summary of various public consultation strategies that Canada may wish to employ.

[44] Supra, note 9. See also Human Genetics Commission, "Public Attitudes to Human Genetic Information: People's Panel Quantitative Study" (2001); Human Genetics Commission, "Report to the Human Genetics Commission on Public Attitudes to the Uses of Human Genetic Information" (2000; People Science and Policy Ltd., "UK Biobank: A Question of Trust, A Consultation Exploring and Addressing Questions of Public Trust" (2002).

[45] Supra, note 1.

[46] Quebec, Commission de l'ethique de la Science et de la Technologie, "The Ethical Issues of Genetic Databases: Towards Democratic and Responsible Regulation," 2003.

Table 6 –
Public Consultation Mechanisms

APPROACH	STRENGTHS	INADEQUACIES
Public opinion surveys	▪ Representative	▪ Superficial coverage of issues
Focus groups	▪ In-depth exploration of reasoning, bases for preferences	▪ Not generalizable
Deliberative consultation	▪ Learning opportunities for lay and expert panels ▪ Interactions with experts, more extensive deliberation	▪ Time and resource intensive ▪ Fewer individuals involved
Stakeholder consultation	▪ Stakeholders' familiarity with issues ▪ Involvement of those with direct benefits or risks	▪ Exclusion of general public
Community consultation	▪ Critical where collectivity is highly valued	▪ Challenge to determine who should represent the community
Web-based consultation	▪ Larger numbers participating ▪ Quick and ongoing information sharing	▪ Individuals selected by technological ability/access/prejudice ▪ Data may not be generalizable
Lay representation on expert committees	▪ Broadened base for considering issues beyond technical considerations	▪ Lay member opinions may be marginalized by expert members

Source: Einseidel, p. 43.

If publics (and other stakeholders) are to participate in a meaningful way in decisions that affect them, they must have access to adequate information upon which their deliberation and the decision-making process can be based. The degree to which publics are knowledgeable about certain topics is variable and is dependent on many factors (Einseidel, p. 10). People may or may not be well informed about a given issue for a number of reasons. They may be passively or actively disinterested: they may be too busy or have no interest or they may decide not to learn about a particular topic. Importantly, disinterest is not necessarily reflective of negative sentiment.

People may be misinformed about an issue due to incorrect or exaggerated information in the popular media. Although the scientific community cannot control the way in which popular media report scientific progress, it can be aware of the importance of the popular media in conveying information to the public. Scientists must consider it their obligation to accurately portray scientific developments to the media and to avoid promoting overly hyped representations of their work. The media should work closely with the scientific community to ensure accuracy and objectivity in their reporting.

Because of the likelihood that publics are currently uninformed or misinformed about genetic technologies, meaningful consultation must depend on engaging and enabling a sufficiently large number of individuals to consider relevant issues once they are provided an appropriate knowledge base.

Canadian funding agencies and/or private partners must accept that public consultation is an essential part of the overall Biobank initiative. Funding requirements for consultation must be budgeted appropriately and considered as part of the overall project strategy (Deschênes and Cardinal, p. 10).

Summary: Consultation, Education and the Role of the Media

- Public education and consultation are necessary first steps if Canada is to be committed to the idea of initiating a large-scale population genetics initiative.
- The consultative process should begin as early as possible in the development of any proposed initiative and should continue for the duration of the initiative.
- The consultative process must be transparent.
- Negative outcomes arising from consultation must be acknowledged and clearly and honestly addressed as they are identified.
- To enhance the value of public consultation, several consultative mechanisms should be employed. A variety of critical, qualitative and quantitative approaches will enable a comprehensive understanding of public (and other stakeholder) sentiment.
- Consultative strategies must incorporate an educational component that will provide a sufficiently detailed knowledge base upon which informed decisions can be based.
- Consultation may be useful in determining how best to convey the concepts of risk and uncertainty to various publics.
- Consultative strategies must be developed to examine the long-term influence of mass media coverage of genetic technologies on biobanking as well as the tools introduced to engage in community-wide consultation to address the issues raised by popular media coverage.

4.2 Recruitment

It is an ethical requirement for the scientific validity and the design of proposed population genetic research to be well founded.[47] Participants in population genetic research must be selected by methods that are supported by scientific, legal and ethical imperatives (Deschênes and Cardinal, p. 10). For example, procedures employed in the recruitment of individuals for research protocols must respect the privacy and confidentiality of the persons (and relatives of those persons) that comprise the potential cohort, and the procedures must be in accordance with privacy legislation within that jurisdiction. In addition, there is a need to ensure that particular segments of the population will not reap unfair benefits nor be unfairly burdened as a result of participating in population genetics research.

The use of personal data to recruit research participants falls under privacy legislation (Deschênes and Cardinal, p. 12). Canadian privacy legislation, at both the federal and provincial levels, generally foresees mechanisms whereby nominative data may be accessed for research without consent if certain conditions are met (for a more detailed discussion, see Section 4.4 of this paper). In the interest of privacy protection, mechanisms for recruitment could be developed wherein participants are notified of the study by provincial health ministries, national bodies responsible for the protection of personal data (e.g., Ombudsman or Privacy Commissioner) or other appropriate institution (e.g., the Canadian Institutes of Health Research). Participants could then be recruited through their general practitioners, medical specialists, or other health care providers. Mechanisms for recruitment that are specifically referable to Canada's health system and the behaviour of patients within the system must be developed.

To ensure that population genetic research will be representative of the overall population, various strategies are recommended. One possibility is the development of a national information campaign to inform the public about population genetics research, including the objectives of the study, the risks and benefits of the research (to the individuals and to society) and proposed safeguards to maintain data security. A national campaign may be coupled with the development of local recruitment strategies to ensure that individuals who are eligible to participate have an opportunity to do so and have information on how to earn more about the project. Local strategies may be developed to endeavour to include individuals from disadvantaged groups (e.g., the low-income population) and individuals

[47] Supra, note 20.

living in rural areas. Communication strategies are key and should also be used to reach those whose first language is not English or French.

Professor Einsiedel notes that the public's willingness to participate in research appears to be mediated by the nature and quality of the information individuals have and how much trust is placed in the medical profession and governing institutions as well as the laws in place to protect them (p. 20, citing Cragg, Ross and Davis, 2000). Individuals are not likely to participate if they are concerned that information obtained about them can be accessed by employers, insurance companies or the police. People are inclined to participate in clinical research if they think their participation will help others and if participation is not too onerous (p. 21).

Summary: Recruitment

- The scientific validity and study design must be well founded prior to recruitment of participants in population genetics research.
- Methods of recruiting participants must be supported on scientific, legal and ethical grounds.
- Mechanisms for recruitment that are specifically referable to Canada's health system and the behaviour of patients within the system must be developed.
- Recruiting strategies must take into account the need to fairly distribute the benefits and burdens of population genetics research to the entire population.
- Meaningful public consultation, genuine debate and educational strategies are likely to maximize willingness to participate in population genetic research.
- The development of a non-coercive national communication and education strategy will help ensure that population genetics research will be representative of and applicable to the entire population.
- Local communication and recruitment strategies should be developed to take into account regional differences and to include participants from all segments of society, including groups that have been traditionally disadvantaged (e.g., the poor, aboriginal communities).
- Participation in population genetic research may be maximized if individuals are assured that they will be recontacted if information is discovered that is relevant to their health status. Enhanced participation based on recontact must be weighed cautiously against the additional burdens in terms of cost, liability, etc. that would arise from such a promise.

- To maximize a population's willingness to participate in population genetics research, assurances should be made that the data collected will be accessible to researchers only for certain clearly defined health-related purposes. For example, participants should be assured that data contained in a population genetic biobank are not accessible to third parties for non-health-related purposes (e.g., law enforcement).

4.3 Privacy and Confidentiality

Privacy

Privacy and confidentiality have frequently been treated as a single concept with interchangeable descriptions. Though closely related, the concepts are distinct. The right to privacy gives an individual the right to control who has access to his or her personal information. For example, individuals have a right to decide whether or not they wish to participate in population genetics research and whether or not to give researchers access to biological samples, associated genetic sequence data derived from the biological samples, personal demographic information, personal health information and genealogical data. Once an individual agrees to participate in research and once information is disclosed to the researcher, the researcher is obligated — both legally and ethically — not to disclose the information to others without permission from the participant. The very nature of DNA makes it uniquely sensitive (Deschênes and Cardinal, p. 27). Concerns exist, for example, that employers, insurance companies or law enforcement agencies might obtain and misuse genetic information (Einseidel, p. 13, citing Canadian Medical Association). Interestingly, most Canadians (more than 90 percent) view genetic information differently from other types of personal information and would like to see stricter rules governing it (Einseidel, p. 13, citing Pollara and Earnscliffe). In particular, 70 percent of Canadians think that legislation to protect the privacy of health information should apply to both the private and public sectors (Einseidel, p. 16, citing Canadian Medical Association).

Privacy, once described simply as "the right to be let alone,"[48] has matured into a concept that is focussed on preservation of an individual's dignity and autonomy;[49] it is viewed as a core human value (for a discussion of definitions, see Yeo, p. 21–23). The notion of privacy includes the right to control the disclosure, dissemination and use of personal information

[48] S. D. Warren and L. D. Brandeis, "The Right to Privacy" (1890), 4 Harv. L. Rev. 193 at 193.

[49] See D. Feldman, Civil Liberties and Human Rights in England and Wales (Oxford: Clarendon, 1993), p. 399 where the author notes: "It is autonomy itself, the freedom to pursue one's own objectives and life-style and to enjoy personal space, which is the fundamental justification for privacy rights."

about oneself. With respect to health information these rights are protected by rules of professional conduct, the common law, the Canadian Charter of Rights and Freedoms,[50] and numerous statutes, both federal and provincial, as well as international treaties, conventions, declaratory statements and ethical guidelines.[51]

Canadian courts have recognized that an individual's "right to security of the person" within the meaning of section 7 of the Canadian Charter of Rights and Freedoms encompasses both the physical and psychological integrity of the individual.[52] It follows that section 7 includes the right to be free from psychological stress that would result from the unauthorized disclosure of one's personal health information.[53] Additionally, section 8 of the Charter, which deems that individuals have the right to be secure against unreasonable search or seizure, has been interpreted to provide protection of an individual's privacy of information. which in this context includes DNA, health and demographic information. This particular right is based on the integrity of the individual and not merely on proprietary interests.[54]

Confidentiality

Patients and their families have a legitimate expectation that confidential health information, including genetic information, will not be disclosed to third parties without permission. This is a fundamental tenet of the fiduciary nature of the doctor–patient relationship.[55] This expectation is reflected in the duty imposed on health care professionals to respect the confidentiality of all information they obtain about their patients. The ethical duty that underlies the legal duty is evident in the Hippocratic Oath and in modern enunciations of it, including the Canadian Medical Association Code of Ethics.[56] Additionally, requirements

[50] Canadian Charter of Rights and Freedoms, Part I of the Constitution Act, 1982, being Schedule B to the Canada Act, 1982 (U.K.), 1982, c. 11.

[51] Ibid. See also Universal Declaration of Human Rights, GA Res. 217, UN Doc. A/810 (1948), Article 12 deals with protection from "arbitrary interference with [one's] privacy, family, home or correspondence" and "attacks upon [one's] honour and reputation." The International Covenant on Civil and Political Rights has a similar provision and is legally binding on its parties, including Canada. Article 12 of that covenant states: "No one shall be subjected to arbitrary or unlawful interference with his privacy, family, home or correspondence, nor to unlawful attacks on his honour and reputation." Other international instruments citing a right to privacy include the United Nations Convention on the Rights of the Child, GA Res. 44/25 (1989), art. 16., and the European Convention on Human Rights (1955), 213 U.N.T.S. 221, art. 8.

[52] R v. Morgentaler, [1988] 1 S.C.R. 30.

[53] Ontario AIDS Society v. Ontario (1995), 25 O.R. (3d) 388, appeal dismissed (1996), 31 O.R. (3d) 798, leave to appeal to the Supreme Court of Canada denied, [1997] S.C.C.A. No. 33.

[54] R. v. Dyment, [1988] 2 S.C.R. 417.

[55] See Re Inquiry into Confidentiality of Health Records in Ontario (1979), 98 D.L.R. (3d) 704 (Ont. C.A.), (1981), 38 N.R. 588 (S.C.C.); Canadian AIDS Society v. Ontario (1995), 25 O.R. (3d) 388 (Gen. Div.); R. v. Osolin (1993), 109 D.L.R. (4th) 478 (S.C.C.); R v. O'Connor (1995), 130 D.L.R. (4th) 235 (S.C.C.), and A. (L.L.) v. B.(A.) (1995), 130 D.L.R. (4th) 422 (S.C.C.). On the issue of the fiduciary nature of the doctor-patient relationship generally, see *McInerney v. MacDonald* (1992), 93 D.L.R. (4th) 415 (S.C.C.).

[56] Canadian Medical Association, "Code of Ethics" (1996), 155 CMAJ 1176A. The physician is obligated to "[r]espect the patient's right to confidentiality except where this right conflicts with [his or her] responsibility to the law, or when

regarding confidentiality have been placed in legislation governing health care facilities, health care sectors and health care professionals. There exists legislation in every province that imposes some obligation on health care providers to maintain patient information as confidential.

There is also a clear common law duty to keep health information confidential.[57] While there is limited jurisprudence in this area, there have been numerous judicial statements that have confirmed the existence of an equitable action for "breach of confidence." As such, there seems little doubt that the handlers of health information can be found liable for the inappropriate use or disclosure of confidential information. For example, the 1990 Alberta Queen's Bench decision of *Hay v. University of Alberta Hospital*, described the duty of confidence as follows:

> *The physician–patient relationship is clothed with confidentiality, a right which may be waived by the patient. Confidentiality is an important attribute of the physician–patient relationship, essential in promoting open communication between physician and patient. The patient may expressly waive this right or, by his actions, be found to have impliedly waived it. Alternatively, an overriding public interest or a statutory direction may justify a physician disclosing information about the patient. In the absence of such circumstances, the right remains and a physician who divulges confidential information could face an action for breach of confidentiality, a possibility which obviously causes physicians some concern.*[58]

The Supreme Court of Canada, in the decision of *McInerney v. MacDonald,* held that certain duties arise from the special nature of the relationship of trust and confidence that exists between doctor and patient. Physicians must "act with utmost good faith and loyalty"[59] in

maintenance of confidentiality would result in a significant risk of substantial harm to others or to the patient if the patient is incompetent; in such cases, take all reasonable steps to inform the patient that confidentiality will be breached."

[57] *Peters-Brown v. Regina District Health Board*,[1995] S.J. No. 60 (Sask. Q.B.).

[58] (1990), 69 D.L.R. (4th) 755 at 757-8.

[59] *Supra*, note 53m McInerney.

their dealings with patients.[60] By extension of these principles, the researcher–participant relationship would likely be determined by Canadian courts to be fiduciary in nature.[61]

Exceptions to the Physician's Duty of Confidence

While physicians and other health care professionals owe a duty of confidence to their patients, there are exceptions, both at common law and in statute. For example, there are numerous legislative exceptions that allow confidential information to be used without the patient's consent.[62] The most controversial exception is the common law "duty to warn."[63] Pursuant to this exception, there may be some circumstances when a physician has a legal and ethical obligation to breach his/her duty of confidentiality in order to protect the health or safety of a third party. The *Tarasoff v. Regents of the University of California* case is the most commonly cited example of this duty.[64] While there are only a few Canadian decisions that explicitly refer to *Tarasoff,*[65] there is a growing body of case law that seems to support the notion that physicians may, in some circumstances, owe a duty to someone who is not a patient.[66] There is also precedent that implies a duty-to-warn exception to the duty of confidentiality.[67] More importantly, many of the emerging provincial health information acts codify a duty-to-warn exception.[68] The Canadian Medical Association recently modified its code of ethics to reflect that the principle of confidentiality is not absolute.[69]

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[60] See also *Norberg v. Wynrib,* (1992) 92 D.L.R. (4th) 449 (S.C.C.); *Henderson v. Johnston* (1956), 5 D.L.R. (2d) 524 (Ont. High Ct.); and *Cox v. College of Optometrists of Ontario* (1988), 65 O.R. 461 (Ont. High Ct.).

[61] Moe Litman and Lorraine Sheremeta, "The Report of the Committee of Inquiry on the Case Involving Dr. Nancy Olivieri: A Fiduciary Law Perspective" (2002), 10 Health L. Rev. 3.

[62] *Health Information Act*, R.S.A. 2000, c. H-5, s.27.

[63] *Ibid*., s. 35(1)(m).

[64] *Tarasoff v. Regents of the University of California*, 551 P. 2d 334 (Cal. 1976).

[65] But see, *Wenden v. Trikha* (1993), 14 C.C.L.T. (2d) 225 (Alta. C.A.).

[66] See, for example, *Pittman Estate v. Bain* (1994), 112 D.L.R. (4th) 257 (Ont. Gen. Div). In this case, the wife of a patient who contracted HIV through a blood transfusion recovered damages from the Canadian Red Cross Society, the hospital and the family doctor for their failure to inform the patient that he had received contaminated blood. This case lends indirect support to the principle that, in exceptional circumstances, a doctor has a duty to warn a patient's partner of the risk of HIV infection, even though the duty involves disclosing confidential information without the patient's consent, and that failure to do so may result in the doctor being held liable in damages. See also *MacPhail v. Desrosiers*, [1998] N.S.J. No. 353 (QL) (C.A.), where the defendant, a physician at an abortion clinic, was held liable for a motor vehicle accident caused by a woman who had just terminated a pregnancy at the clinic.

[67] *Supra*, note 53, McInerney at 154:

> *The right [of confidentiality] is absolute unless there is some paramount reason that overrides it. For example, there may be cases in which reasons connected with the safety of individuals or of the public, physical or moral, would be sufficiently cogent to supersede or qualify the obligation prima facie imposed by the confidential relation.*

[68] *Supra,* note 60, Section 35(1)(m) allows the disclosure of confidential health information "to any person if the custodian believes, on reasonable grounds, that the disclosure will avert or minimize an imminent danger to the health or safety of any person."

[69] See Canadian Medical Association, *Code of Ethics* (Ottawa: CMA, 1996), which allows a breach of patient confidentiality if there is "a significant risk of substantial harm to others."

The duty to warn is relevant in the context of the collection and use of human biological samples. It has been suggested that there may exist circumstances when a physician has a legal obligation to warn a family member of a patient of a potential health risk revealed through genetic testing of the patient. Because a patient's genetic information may be relevant to a relative (e.g., for decisions about reproduction or treatment options), it has been argued that there may be a "duty to warn" a family member about a given genetic risk or condition.[70] The communication of information to family members at high risk for serious harm without the consent of the research participant should be contemplated **only** when all attempts to elicit voluntary cooperation and communication of the information by the tested individual to his or her relatives have failed.[71] Despite this trend toward recognition of a duty to warn, it is still very unclear when it would apply. For example, there is no consensus on how severe the risk of harm to the third party must be in order to trigger the duty.

Privacy Legislation in Canada

In Canada, there are numerous statutes — both federal and provincial — that are relevant to health information. For example, Table 7 demonstrates a variety of key statutes that are relevant to the privacy and confidentiality of health information in Alberta. The complexity of the legislative framework is magnified when one considers that the situation is similar in most Canadian provinces.

Table 7 –
Examples of Alberta Statutes Relative to Privacy and Confidentiality of Health Information

Health information	▪ *Health Information Act*, Bill 40, 2nd Sess., 24th Leg., Alberta, 1999.
Freedom of information and protection of privacy	▪ *Freedom of Information and Protection of Privacy Act*, S.A. 1994, c. F-18.5, s. 22.03.
Public health	▪ Public Health Act, S.A. 1984, c. P-27.1.
Health administration	▪ *Health Care Insurance Act*, R.S.A. 1980, c.A-24, s. 13. ▪ *Health Care Insurance Regulation*, AR 216/81, s. 23. ▪ *Hospitals Act*, R.S.A. 1980, c. H-11, s. 40.
Cancer	▪ *Cancer Programs Act*, R.S.A. 1980, c. C-1, Part 1.1. ▪ *Cancer Programs Regulation*, AR 242/98

[70] For relevant jurisprudence, see *Tarasoff v. Regents of University of California*, 551 P. 2d 334 (Cal., 1976) and *W. v. Egdell*, [1990] 2 W.L.R. 471 (C.A.).

[71] B. Knoppers and R. Chadwick, "The Human Genome Project: Under an International Ethical Microscope" (1995), 265 Science 2033 at 2033.

Freedom of information legislation applies to personal information in the **public sector** and now exists federally, provincially and in the territories. There is new federal privacy legislation that concerns the collection, use and disclosure of personal information in the **private sector**. Numerous provinces are currently in the process of drafting similar privacy legislation. The effect of "substantially similar" provincial legislation would be to pre-empt the operation of the federal statute in the provinces with such legislation. Numerous provinces now have specific legislation governing the privacy of health information.

Table 8 –
Relative Coverage of Federal and Provincial Privacy Statutes (or Bills) in Canada

Jurisdiction	Right of privacy	Criminal law	Tort	Clinical records	Registries statistics	Freedom of information, public	Protection of personal information, health	Protection of personal information, private
Canada	4	4			4	4		4
British Columbia			4	4	4	4		(4)
Alberta				4	4	4	4	(4)
Saskatchewan			4	4	4	4	(4)	
Manitoba			4	4	4	4	4	(4)
Ontario				4	4	4	(4)	(4)
Quebec	4			4	4	4		4
Nova Scotia				4	4	4		
New Brunswick				4	4	4		
Prince Edward Island				4	4	4		
Newfoundland and Labrador			4		4	4		
Yukon					4	4		
Northwest Territories					4	4		
Nunavut					4	4		

4 denotes legislation in force.
(4) denotes pending bill.

Source: Kosseim, slide 9.

Appendix C provides a province-by-province listing of statutes and bills.

Federal Personal Information Protection and Electronic Documents Act (PIPEDA)

The federal Personal Information Protection and Electronic Documents Act (PIPEDA)[72] imposes mandatory standards for the collection, use and disclosure of personal information, including health information, in the **private sector**. The stated purpose of PIPEDA is:

> *to establish, in an era in which technology increasingly facilitates the circulation and exchange of information, rules to govern the collection, use and disclosure of personal information in a manner that recognizes the right of privacy of individuals with respect to their personal information and the need of organizations to collect, use or disclose personal information for purposes that a reasonable person would consider appropriate in the circumstances.*[73]

Schedule 1 of PIPEDA specifically incorporates the principles enunciated in the Canadian Standards Association Model Code for the Protection of Personal Information into the Act. In summary, these principles are summarized in Table 9:

Table 9 –
Fair Information Principles as Set out in the National Standard of Canada Titled "Model Code for the Protection of Personal Information," CAN/CSA-Q830-96

PRINCIPLE	DESCRIPTION
Accountability	An organization is responsible for personal information under its control and shall designate an individual or individuals who are accountable for the organization's compliance with the following principles.
Identifying purposes	The purposes for which personal information is collected shall be identified by the organization at or before the time the information is collected.
Consent	The knowledge and consent of the individual are required for the collection, use or disclosure of personal information, except where inappropriate.
Limiting collection	The collection of personal information shall be limited to what is necessary for the purposes identified by the organization. Information shall be collected by fair and lawful means.
Limiting use, disclosure and retention	Personal information shall not be used or disclosed for purposes other than those for which it was collected, except with the consent of the individual or as required by law. Personal information shall be retained only as long as necessary for the fulfilment of those purposes.
Accuracy	Personal information shall be as accurate, complete and up-to-date as is necessary for the purposes for which it is to be used.
Safeguards	Personal information shall be protected by security safeguards appropriate to the sensitivity of the information.
Openness	An organization shall make readily available to individuals specific information about its policies and practices relating to the management of personal information.

[72] *Personal Information Protection and Electronic Documents Act,* R.S.C. 2000, c.5 (PIPEDA).

[73] *Ibid*, s. 3.

PRINCIPLE	DESCRIPTION
Individual access	Upon request, an individual shall be informed of the existence, use and disclosure of his or her personal information and shall be given access to that information. An individual shall be able to challenge the accuracy and completeness of the information and have it amended as appropriate.
Challenging compliance	An individual shall be able to address a challenge concerning compliance with the above principles to the designated individual or individuals accountable for the organization's compliance.

As of January 1, 2002, PIPEDA became applicable to personal health information but until January 1, 2004, this remained limited to personal health information in the federally regulated private sector and to cross-border disclosures of such information. After January 1, 2004, PIPEDA also applies to the health information in the private sector. Where a province enacts its own legislation covering the private sector and the provincial legislation is substantially similar to PIPEDA, the provincial law applies.

Specifically, PIPEDA contains provisions that contemplate the use and disclosure of personal information for research purposes.[74] Personal information may be used or disclosed for research without the subject's knowledge or consent for purposes that cannot be achieved without using or disclosing the information, where it is impracticable to obtain consent, and where the organization informs the Privacy Commissioner prior to the use or disclosure of information. The information must, however, be used in a way that maintains its confidentiality.

PIPEDA has been criticized for failing to recognize the nature of health information and the special aims of health care provision. It also fails to recognize the pre-existing legal and ethical obligations that physicians and researchers owe to patients and research subjects. Additionally, it has been suggested that the fair information principles underlying the Act do not necessarily accord with the norms of health care provision.

Provincial Legislation for the Protection of Personal Information in the Private Sector

As noted above, the federal PIPEDA applies to all personal health information collected, used or disclosed during the course of commercial activity as of January 1, 2004. If a province enacts its own legislation that is "substantially similar" to PIPEDA, the provincial legislation will apply within the province. Quebec has had legislation in place since 1993.[75]

[74] *Supra*, note 70 (PIPEDA), ss. 7(2)(c) and 7(3)(f).

[75] *An Act Respecting the* Protection *of Personal Information in the Private Sector*, R.S.Q., c. P-39.1.

Alberta (Bill 44[76]) and British Columbia (Bill 38[77]) have drafted legislation that, if passed, will apply to private sector transactions.

Many questions remain about the interaction of the federal and provincial statutes with respect to biobanking. Which act will properly apply? Will the provincial statutes be deemed "substantially similar" to the federal legislation? The many privacy laws that exist, though conceptually similar, are not identical. The degree to which the laws will be harmonized is yet to be seen. At present, it appears that navigating the existing and developing legislative framework in the context of biobanking will be a difficult challenge. This is particularly true considering that "biobanking" in the context of developing a population genetics research resource is neither strictly public nor strictly private and it is not a typical commercial venture.

Provincial Privacy Legislation Concerning Health Information

A number of Canadian provinces including Alberta, Manitoba and Saskatchewan have enacted specific legislation concerning the privacy of health information. Such legislation aims to:

- establish mechanisms to protect the privacy of individuals with respect to their health information and to protect the confidentiality of that information
- enable health information to be shared and accessed
- facilitate the provision of health services and management of the health system
- prescribe rules for the collection, use and disclosure of health information
- provide individuals with a right of access to health information about themselves
- provide individuals with a right to request correction or amendment of health information about themselves
- establish strong and effective remedies for contraventions of the statutes
- provide for independent reviews of decisions made by custodians or trustees pursuant to the acts and the resolution of complaints.

In general, health information legislation accomplishes these objectives by:

- placing clear obligations on the "custodians" of health information, with respect to "personally identifying" health information

[76] Bill 44, *Personal Information Protection Act*, 3rd Sess., 25th Parl., Alberta, 2003 (First Reading 14 May 2003).

[77] Bill 38, *Personal Information Protection Act*, 4th Sess., 37th Parl., British Columbia, 2003 (First Reading 30 April 2003).

- setting rules governing the collection, use, storage, disclosure, retention, disposal and destruction of personal health information, including biologic samples (see, for example, *Alberta Health Information Act* s. 1(1)(i)(iii))
- demanding that health information can be used and/or disclosed only for the purpose or purposes for which it was collected or for a consistent purpose
- permitting personal health information to be used and/or disclosed for research purposes with or without the consent of the individual to whom the information pertains if certain criteria are met (see, for example, *Alberta Health Information Act* s. 48–56).

Careful analysis of the federal and provincial privacy statutes relative to biobanking is required. Biobanking is a complex subject matter that is not obviously and neatly either public or private. The federal jurisdiction over trade and commerce and the provincial jurisdiction over health further complicate the matter. The complexity of the legislative web may compel consideration of biobank-specific legislation that could address the complex privacy issues in a more direct and coherent way.

Table 10 summarizes the public opinion data relating to issues of privacy.

Table 10 –
Summary of Public Opinion Data Relating to Issues of Privacy

PUBLIC CONCERNS	
Canada	▪ Nearly 90% of Canadians regard genetic information as being different from other types of personal information and want to see stricter rules for governing access to such information (Einseidel, p. 13, citing Pollara and Earnscliffe, 2001) ▪ Two-thirds of Canadians feel that genetic information is "most private and confidential" and they do not want others to have access without consent (Einseidel, p. 13, citing Canadian Medical Association, 2000) ▪ Concerns exist regarding donor anonymity, records being used for research purposes and the possibility of employers and insurance companies obtaining and misusing the information (Einseidel, p. 13, citing Canadian Medical Association, 2000). ▪ About 78% of Canadians polled agree that doctors **should** have access to genetic information for purposes of diagnosis and therapy; 60 % think that provincial health ministries should **not** have access to genetic information; 87% think that private insurance companies should **not** have access to genetic information; and 63% think that the police should have access to genetic information to solve crimes (Einseidel, p. 14, citing Einsiedel, forthcoming). ▪ About 7 in 10 Canadians think that legislation to protect the privacy of health information should apply to both public and private sectors (Einseidel, p. 16, citing Canadian Medical Association, 2000). ▪ Most people (76%) presume that genetic information produced in any test is preserved, though they tend to think more of the lab's premises than a data bank. Almost 60% think that is true of the actual sample of blood or saliva as well (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 10). ▪ Absent arguments about the benefits that might be derived from the use of personal genetic information, most Canadians default toward the strict protection of genetic privacy (Pollara and Earnscliffe, *Genetic Privacy Issues,* p. 14). ▪ Privacy is not the only priority Canadians consider. They highly value health and medical uses of genetic information. This is particularly true with respect to the development of cures for genetically based diseases. Canadians are generally quite open to research uses of genetic information (Pollara and Earnscliffe, *Genetic Privacy Issues,* p. 14).

PUBLIC CONCERNS	
	▪ Canadians are divided about whether genetic information is fundamentally different from health information, but they do expect access to be more strictly regulated than other for medical information (Pollara and Earnscliffe, *Genetic Privacy Issues,* p. 9). ▪ Canadians express widely differing views in terms of comfort with different groups/individuals having access to genetic information. They express a high level of comfort with doctors and medical researchers having access and a low level of comfort with insurance companies, employers and governments including provincial health departments having access to this information (Pollara and Earnscliffe, *Genetic Privacy Issues,* p. 107).
United Kingdom	▪ Concerns about misuse of genetic information by employers and insurers are adequately addressed if information is provided about why the information is useful, how it will be used and the safeguards that are in place to guard against unauthorized access (Einseidel, p. 13, citing Human Genetics Commission, 2002). ▪
United States	▪ As a result of increasing concern among its constituent publics, the U.S. Congress introduced a rule requiring researchers using tissue banks to obtain consent when using individually identifying patient information (Einseidel, p. 12, citing Uraneck, 2001). ▪ Some 85% of Americans surveyed in 1995 were "very concerned" or "somewhat concerned" that insurers and employers would gain access to and misuse genetic information (Einseidel, p. 15, citing Uraneck, 2001) ▪ Fewer than 1 in 5 Americans considered the use of medical records for medical research without permission to be very acceptable; fewer than 1 in 3 thought it unacceptable (Einseidel, p. 15, citing Uraneck, 2001).
PROFESSIONAL CONCERNS	
	▪ There is disagreement among professionals about the conditions for confidentiality and the viability of technical solutions to provide answers (Einseidel, p. 30).

Summary: Privacy and Confidentiality

- Numerous factors make the issues surrounding privacy and confidentiality particularly challenging, including the rapid developments in information technology and genetics and the ongoing tension between the desire to do beneficial health research and the need to protect personal health information.

Privacy

- At present, it appears that there exists no coherent legal framework within which to appropriately address health-related privacy issues that are relevant to biobanking.
- It is essential to gain an understanding of the interaction between federal and provincial privacy statutes relative to biobanking. Jurisdictional issues between the federal power over trade and commerce and the provincial power over health must be reconciled.
- The existing legislative framework is complex and may compel consideration of biobank-specific legislation that could address the complex privacy issues.
- There is an need to develop encryption, anonymization and data sharing methods to ensure security of data according to identified needs.[78]

[78] *Supra*, note 1. See Appendix B of this paper.

Confidentiality

- Physicians have a clear, common law duty, including a fiduciary duty, to keep patient information confidential (a duty that has been modified by privacy legislation).
- There are a number of exceptions to the duty of confidentiality, such as those set out in health information legislation. The most controversial exception, however, is the "duty to warn" third parties. Though emerging case law and legislation lend support to the existence of the duty, its scope remains uncertain.

4.4 Informed Consent and Communication of Research Results

Individual Consent to Research

In Canada, to be valid, consent to medical treatment must be specifically referable to the treatment to be given, and it must be given voluntarily by a patient who has capacity and is informed. If any one requirement fails to be met, the consent is void. The consent requirements in the context of clinical research are more onerous than for medical treatment. The *Nuremberg Code* and the *Declaration of Helsinki* and the Tri-Council Policy Statement (TCPS) require that participants in medical research provide voluntary informed consent prior to participating in medical research. Importantly, informed consent must be viewed as an ongoing process and participants must be given adequate opportunities to discuss and contemplate continuing participation.

Canadian courts have determined that research subjects are entitled to a "full and frank disclosure of all the facts, probabilities and opinions which a reasonable man might be expected to consider before giving his consent."[79] This obligation requires that researchers must provide information about potential risks (regardless of how remote) as well as all material information about the research protocol. In the context of clinical genetics research, study participants must be apprised of the potential for commercialization of research findings, the mechanisms used to protect confidentiality of sensitive information, the potential impact of participation on one's insurability, their entitlement to withdraw from participation in the study at any time and how (or if) the study results will be made available to the participant or the participant's physician. Appendix D provides a list, extracted from the TCPS, of the types of information that should be conveyed to potential research subjects.

[79] *Halushka v. University of Saskatchewan* (1965), 53 D.L.R. (2d) 436 at 443-44.

Retrospective Research Involving Previously Existing Collections

With respect to retrospective research on stored human tissues and associated data, health information legislation or other privacy laws typically provide an exception to the general rule that informed consent is required for research using personally identifiable health information or tissue. For example, Alberta's *Health Information Act* allows researchers to access identifiable health information, including tissue, without consent, under specific circumstances with research ethics board approval.[80] In reviewing the research proposal, the research ethics board must consider whether, among other things, the proposed research is of sufficient importance to outweigh the public interest in protecting privacy, and whether obtaining consent is unreasonable, impractical or not feasible.

Despite this legislative weakening of the informed consent requirement in the context of retrospective research, Canadian law and developing international norms strongly support the need for individual consent to prospectively collect and store DNA samples and personal information in a population genetics biobank.

Prospective Research

In Canada, strict application of existing law and policy in the context of biobanking would demand that consent be obtained at the time a sample is collected and again for each new use of an identifiable DNA sample or associated data in a biobank (Deschênes and Cardinal, p. 35, citing Caulfield and Outerbridge). This requirement is difficult to fulfil in the context of biobank research where potential future uses are not known. The specific degree of detail to fulfil informed consent requirements in the context of population genetic research remains speculative. The Human Genetics Commission has stated that:

> *[t]he difficulties involved in tracing and securing reconsent for different forms of medical research may make obtaining fresh consent impractical and would seriously limit the usefulness of large-scale population databases.*[81]

[80] *Supra*, note 60 (*Health Information Act*), ss. 48-55.

[81] *Supra*, note 9.

Similarly, using only anonymous samples or utilizing a process to irreversibly anonymize samples would limit the usefulness of the samples and resulting data. The value of biobanks for future research is greatly enhanced if mechanisms are used that protect the identity of research participants while maintaining links to health information and other data that can updated over time (Deschênes and Cardinal, p. 28).

Commentators have questioned the appropriateness of the legal norms governing consent in this setting. Caulfield et al. cite cogent reasons why the traditional norms may properly be considered too onerous in the context of biobanks and population genetic research. [82] The reasons can be summarized as follows:

- The full spectrum of biotech research is difficult, if not impossible to predict at the time consent is obtained for sample collection.
- The collection of DNA samples (typically through venipuncture or buccal swab) involves little risk to the individual participant.
- Biobank research is not likely to provide information that is of direct clinical relevance to individual participants.
- The value of individual samples in a biobank is marginal; the value resides in the aggregate collection and the ability to analyze in a multitude of ways.
- Repeat requests for individual consent from biobank participants is burdensome to researchers and to participants and may be a disincentive to participation.

Various policy options have been suggested that alter the informed consent requirement to a greater or lesser degree. Table 11 provides a comparison of models of consent, including fully informed consent, “authorization model,” blanket consent, presumed consent and waiver of consent.

[82] *Supra*, note 18.

Table 11 –
Comparison of Existing Consent Models

CONSENT MODEL	FEATURES
Informed consent	▪ Informed consent is required for the initial collection of the biological sample and for each subsequent new use in a research protocol. ▪ May be overly restrictive with respect to future research. ▪ Recontacting of patients/participants is onerous and may not be desirable in certain circumstances (e.g., when patient/research participant is deceased).
Authorization model (See Deschênes and Cardinal, pp. 16–17; Yeo, p. 49)	▪ Informed consent is required for the initial collection of the biological sample. ▪ Subsequent research is authorized (or not) by the research participant at the time of the initial sample collection. ▪ Individuals can specify specific permitted or excluded uses of their biological material and associated data and can specify the degree of subsequent decision-making authority they want to maintain. ▪ There is a possibility that individuals can opt for a general "blanket consent" but is never presumed by researchers. ▪ This model strikes a reasonable balance that is supportive of individual autonomy and of genetic research and is supported by the TCPS (Article 8.6). ▪ This model accords with public opinion.
Blanket consent (Deschênes and Cardinal, p. 14)	▪ One-time consent, wherein a research participant consents to "research in general." ▪ Too general to be afforded much legal weight, a person cannot be held to have consented to that to which he has given no consideration. ▪ Not supported by public opinion: the public has the expectation that rules governing access to genetic information should be strengthened. ▪ Some argue that the principle of autonomy supports the right of research participants to give blanket consent to research; others argue that blanket consent is no consent at all.
Presumed consent	▪ Because a patient or research participant donated a biologic sample for research and because only a relatively small proportion of individuals would want to reconsent, consent to future uses can be presumed. ▪ Presumed consent is problematic because of the degree to which it undermines respect for the individual participant and the right of the individual to make informed decisions about matters involving him or her. ▪ Not supported by public opinion. ▪ May be a justifiable presumption to permit access to and use of existing collections of human biologic materials that would otherwise be of little value.
Waiver	▪ Requirement for obtaining informed consent from individuals to whom individually identifying data or samples refer can be waived by a research ethics board if (among other things) the research poses no more than minimal risk to the subjects (see TCPS, s 2.1(c); and see *Alberta Health Information Act*, ss. 48–55). ▪ Not supported by public opinion. ▪ Some feel a waiver is justifiable to permit access to and use of existing collections of human biologic materials that would otherwise be of little value.

With respect to the various options described in Table 11, the authorization model appears to be the best option for large-scale population genetic research. Implementation of an authorization model of consent would, however, require changes in the current regulatory and legislative environment.

Research Involving Children

The proposed Canadian birth cohort study envisions the enrolment of infants in a longitudinal population genetic research protocol. In Quebec, pursuant to the Civil Code,[83] an incompetent minor may participate in research with parental consent, provided that the minor does not object and there is no serious risk. In that province, population genetic research involving children would require approval by a research ethics board constituted or designated by the Minister of Health and Social Services. The proposed experiment "must have the potential to produce a benefit to the health of the person concerned or, if it is conducted on a group, to the health of the persons in the same age group or having the same illness or handicap as the persons submitted to the experiment." In other provinces, the legality of research involving children is unclear.[84] Further consideration of this issue is required.

The TCPS anticipates that parents, as authorized representatives of children, may consent to research on behalf of the minor if the research does not expose the minor to more than minimal risks without the potential for direct benefits. [85] Infants and children may be considered to participate in research when the research question can be addressed using individuals within the identified group(s) only. In the case of population genetic research involving children, the argument has been made that a lifelong birth cohort is necessary to study the interaction between human genes and environmental factors. If academic debate confirms the scientific merit of this approach and if the law permits research on children in this context, ethics would require that the parent(s) or guardian(s) consent as the "authorized representative" of the minor.

Population Consent

Although the notion of population consent is contentious and difficult, if not impossible to implement, it is important to inform and consult the population and to consider public opinion (Deschênes and Cardinal, p. 8). Canadians ought to be consulted as to whether and in what circumstances they would consider participating in a national biobank initiative. Appropriate forums for discussion must be established and supported by the planners and funders of large-scale population genetic initiatives (see Section 4.1 above).

[83] *Civil Code of Quebec*, S.Q., 1991, c. 64, s. 21.

[84] Ellen I. Picard and Gerald B. Robertson, *Legal Liability of Doctors and Hospitals in Canada* (Scarborough: Carswell, 1996), 90-2; Kathleen Cranley Glass and Trudo Lemmens, "Research Involving Humans," in *Canadian Health Law and Policy*, edited by Jocelyn Downie, Timothy Caulfield and Colleen Flood, 2nd ed., pp. 481–90 (Markham: Butterworths, 2002), p. 458

[85] *Supra*, note 20, art. 2.6.

Public Opinion Data

The most thorough research on the issue of consent has been performed in the United Kingdom. Laws governing consent in Canada are more onerous than those in the United Kingdom and health law jurisprudence in this country places greater emphasis on the principle of autonomy.[86] Though these data provide a useful starting point, similar studies must be performed in Canada (Einseidel, p. 44).

From the data on public opinion presented by Einsiedel, it is clear that the publics feel strongly about the value of informed consent for population genetic research. Unanimously, all groups studied (general public, patient groups, religious leaders, special interest groups and professionals) held the opinion that informed consent is **crucial** (Einseidel, pp. 11, 27). There is particular concern is that that third parties (including employers and insurance companies, law enforcement agencies) may obtain access to biological samples and/or associated data.

Strategies that have been suggested for dealing with informed consent in the context of population genetic research, including waiver of consent and blanket consent, appear to be inconsistent with public opinion. Given the need to foster and maintain public research in the area of genetic research and the growing concerns about privacy and confidentiality, it is necessary to glean insight into the attitudes of Canadian publics toward biobanks.

[86] Timothy Caufield and Tim Outerbridge, "DNA Databanks, Public Opinion and the Law" (2002) 25 Clin. Invest. Med. 252.

Table 12 –
Summary of Public Opinion Data Relating to Issues of Consent

PUBLIC CONCERNS	
Canada	▪ Fewer than 50% of Canadians think that health information could be released to governments and researchers without consent, provided that identifying data have been removed (Einseidel, p. 12, citing Canadian Medical Association 2000). ▪ In focus groups, most people after discussion are comfortable with researchers accessing biobanks for a variety of studies, including those that were not contemplated at the time of consultation (Pollara and Earnscliffe, *Genetic Privacy Issues*). ▪ The consistent provisos are that people provide informed consent (though only at first instance, not at each different research use) and that their identity be masked or stripped away (although large numbers of people would accept researchers having access to their identity if it furthered the cause of medical research and was not used inappropriately) (Pollara and Earnscliffe, *Genetic Privacy Issues*).
United Kingdom	▪ About 9 in 10 people surveyed think that consent should be required prior to blood or tissue being used for genetic testing (Einseidel, p. 12, citing Human Genetics Commission, 2001). ▪ Almost 9 in 10 feel that fresh consent should be obtained before research is permitted on existing samples (Einseidel, p. 12, citing Human Genetics Commission, 2001). ▪ Some members of the public see the fact that no feedback will be given as problematic and something that could pose a barrier to participation (Einseidel, p. 13, citing *People, Science and Policy*, 2002). ▪ Donors should have the right to feedback on anything that emerged from their own samples (Einseidel, p. 13, citing Porter, 2000). ▪ Some members of the public understand and accept that feedback would not be provided to individuals. Some think this would be problematic and could pose a barrier to participation (Einseidel, p. 13, citing *People, Science and Policy*, 2002). ▪ There is interest in receiving general information on discoveries or developments made from the research (Einseidel, p. 13, citing Porter, 2000).
United States	▪ More than 8 in 10 Americans consider the use of patient records for medical research without prior permission to be unacceptable (Einseidel, p. 11, citing Institute for Health Care Research and Policy, 1999). ▪ While comfort levels increase if the information is not linked to individual patients, 1in 3 consider use of such information “not at all acceptable” without patient consent (Einseidel, p. 11, citing Institute for Health Care Research and Policy, 1999). ▪ Most people would not object to research being performed on samples to which demographic information or medical history was linked (Einseidel, p. 13, citing NBAC, 2000). ▪ Individuals who are to be notified in the event that medically helpful information is discovered about them tend not to object to the idea of research in which demographic data are linked to stored tissue samples; they are only slightly more concerned about links to medical histories (Einseidel, p. 13, citing NBAC, 2000).
CONCERNS OF PROFESSIONALS	
United Kingdom	▪ Health care professionals are generally adamant that consent be obtained at point of collection and for subsequent uses (Einseidel, p. 27, citing Hapgood, 2001).
Canada	▪ Researchers admit that information about ownership, control, storage conditions and sharing of samples between researchers is often not shared with tissue donors (56% did not specify ownership information; only 15% specified storage duration; about 60% admit to sharing samples with other researchers) (Einseidel, p. 27, citing Verhoef et al., 1995).
United States	▪ Two-thirds of allied health professionals in the U.S. support autonomy in situations where subjects choose not to learn the results of genetic testing; 29% would put limits on autonomy when tested individuals refuse to tell at-risk relatives of the results (Lapham et al., 1997). ▪ Some 95% of those polled think that permission should be obtained before the release of medical records to a national database; 93% believe that researchers should obtain permission to study an individual’s genetic information (Gallup, 2000).
Iceland	▪ The Icelandic Medical Association warned the Icelandic government that the interests of patients were jeopardized by unacceptable arrangements concerning informed consent (Einseidel, p. 34, citing Icelandic Medical Association, 2000).
United Kingdom	▪ Health care professionals feel that the concepts of risk and commercial gain are inadequately explained in patient information sheets and frequently treat DNA collection from blood or tissue as incidental to the research (Einseidel, p. 27, citing Rigby, 2001).

Summary: Informed Consent

- It appears that individual informed consent has become the *de facto* norm for prospective population genetic research.
- Obtaining informed consent is problematic in the context of large-scale population genetic biobanks to the extent that the future research uses of the tissue and/or data to be derived therefrom are unknown.
- Informed consent must be viewed as an ongoing process and it must be reaffirmed if significant changes are made to the protocol or banking conditions.
- Implementation of large-scale population genetic research requires a reassessment of the current normative framework governing informed consent.
- Legislating an "authorization model" of informed consent is worthy of consideration in the specific context of prospective population genetic research.
- Participants in population genetic research must be able to withdraw their consent to participate. Procedures must be in place to facilitate withdrawal of all data that are personally identifying or that are capable of being personally identifying.
- If Canada wishes to embark on a large-scale population genetic initiative involving minors, the legality of consent by authorized representatives in this context must be established.
- Details of feedback must be determined at the outset of population genetic research and communicated to research participants.
- Out of respect for the subject population, research results must be publicly shared; results should be shared in a timely and diligent manner.
- The use of previously existing collections of human biological materials for genetic research without individual consent is possible in limited circumstances.
- Individual participants must be aware of the risk (if any) that law enforcement agencies (or others) may obtain access to data and samples before giving consent and before samples are taken.

4.5 Commercialization

Despite calls for public consultation and deliberation over whether or not Canada should embark on a large-scale population genetic biobank initiative, the presumption is that the current proposal of the Canadian Institutes of Health or a similar initiative will inevitably proceed. Given this reality, we are obliged to consider how to best to manage real and perceived conflicts of interest and how the effects of commercialization and the rapid dissemination of clinically useful genetic innovations can be facilitated in a manner that best accords with the public interest and results in the greatest possible benefit to society.

Increasingly the distinction between the private and public sectors is blurred in the area of biomedical research. All of the authors of the previously commissioned papers highlight the issue of commercialization as being potentially problematic in the context of population genetic research. Specific concerns include the following:

- commodification of the human body
- ownership of human biological materials and associated data
- academic secrecy
- skewing of the research agenda from basic research to the development of commercializable end products
- premature implementation of new technologies in the marketplace (i.e., before the clinical, ethical, legal and social issues have been appropriately considered)
- potential adverse effects of patents (and other forms of intellectual property) on patient access to new technologies.

Private industry will play a critical role in the translation of the information derived from the Human Genome Project into tangible products and procedures that will benefit individuals and society. The private sector funds a great deal of research and relieves academic researchers of mundane and repetitive research. It performs the tightly controlled independent studies that are required for drug and medical device approval processes. Although private industry contributes more to genetic research in terms of funding than do governments, it is recognized that government research frequently gives industry a jump-start on the path to commercialization. The role of public sector funding in the commercial process must not be marginalized.

The public sector's increasing dependence on private enterprise to fund and participate in basic, translational and clinical research provides new opportunities for conflicts of interest to arise (Deschênes and Cardinal, p. 26; Yeo, p. 18; Einseidel, p.17). There are concerns, for example, that private industry will place inappropriate limitations on the academic freedom of publicly funded clinical researchers, that researchers will become less collaborative and will be less inclined to share data with colleagues and the wider research community, that the focus of biomedical research will be skewed away from basic research to what is likely to be commercializable, and that the growing dependence of academia on industrial partners is likely to have an adverse impact on public trust. The related and ongoing debate over the ethical appropriateness of gene patenting and stem cell patenting has not abated. There is a real risk that overemphasis on commercial efforts will undermine science, scientists, the fruits of research and the medical profession.

Public opinion data suggest that, although the public generally supports biotechnology and the development of the biotech sector, there are serious reservations about certain aspects of commercialization and ownership of human genetic material (Einseidel, p. 17). For example, among Canadians there is a deep resistance to the idea of biobanks selling genetic data to others doing research, even with consent (Pollara and Earnscliffe, *Genetic Privacy Issues*, 2003). It is thought that this implies more a judgment against the role of profit in association with health care than a considered decision about the particular circumstances. Public opinion data from Canada and abroad suggests that the public generally lacks trust in corporate responsibility in the biotechnology field and that it trusts researchers less if they are collaborating with industry. The issue of trust is particularly relevant and must be addressed if a large-scale population genetic initiative is to be commenced.

Table 13 –
Key Issues That Have Arisen over Commercial Involvement in Biobank Projects

Iceland	Serious concerns have been raised over the grant of an **exclusive licence** to deCODE Genetics to exploit the Health Sector Database for profit. This situation highlights the clear need to clarify, at the outset, the purpose and principles that underlie any large-scale population genetic initiative. Public consultation is essential in defining the purpose and principles if the project is to inspire trust in the public.
UK Biobank	Consultation in the United Kingdom revealed public concerns over **any** commercial involvement in Biobank. People generally feel that databases should not be owned by commercial interests and that products developed through the initiative should be publicly owned (Einseidel, pp. 15–16). This situation highlights the need to develop liberal access mechanisms for researchers, sharing of research results and transparent benefit-sharing mechanisms to ensure direct and obvious public benefit.
DNA Sciences Inc.	The "Gene Trust" was established by DNA Sciences through the solicitation of volunteers over the Internet. A sample repository of more than 10 000 samples and associated data was established. It was recently sold to Genaissance Pharmaceuticals in a deal whereby DNA Sciences sold substantially all of its assets to Genaissance to avoid bankruptcy. This situation highlights a critical need to examine mechanisms in corporate law that could be used to protect donor participants (e.g., legal trust).
Tonga	The Tongans effectively opposed the creation of a biobank and patenting of their genetic resources by Autogen because they felt that the benefits promised (including free drugs and royalties) were insufficient in light of what Autogen stood to gain (Deschênes and Cardinal, p. 24). This situation highlights the need to include the public in negotiations over benefit sharing to make sure that they feel a fair deal has been reached. It also reflects a need to consider tying royalties to future commercial success so that a population would gain proportionately and appropriately from a future blockbuster product.

Deschênes and Cardinal (p. 26–27) aptly point out that the creation and exploitation of population genetic biobanks in Canada requires careful consideration of the following questions:

- Can the commercialization of products and services developed from population genetic research be simultaneously promoted and aligned with the best interests of society?
- Who can or should own or control a population genetic biobank?
- How should the interests of the Canadian population be represented in any commercial agreements that flow from biobanking?
- Is there an emerging legal obligation that would require Canada to incorporate benefit sharing into population genetic initiatives?
- How might benefit-sharing arrangements be implemented in the Canadian context?

Where populations or communities contribute to research projects and where profits ultimately accrue to a commercial entity (or entities), it is appropriate to consider whether and how profits and other benefits should be shared with the participant community. Benefit-sharing arrangements, if carefully considered, provide a key mechanism for establishing and maintaining public trust in the context of population genetic initiatives. It is very clearly ethically mandated and there is evidence of an emerging legal requirement in international

law.[87] The HUGO Ethics Committee Statement on Benefit Sharing discloses potential mechanisms that may be used to effect benefit sharing between sponsor companies and communities that participate in population genetic research.

Benefit sharing should not be viewed as contrary to the existing intellectual property regime nor as a mechanism to curb commercial involvement in genetic research. Rather, benefit sharing should be considered as a mechanism (or rather a spectrum of mechanisms) to balance the commercial interests with those of research participants in a way that is both respectful and reflective of the relative contributions to the research endeavour.

Table 14 provides an overview of types of monetary and non-monetary benefits that may be included in benefit-sharing agreements.

Table 14 –
Monetary and Non-monetary Benefits That May Be Incorporated into Benefit-sharing Agreements

MONETARY BENEFITS	NON-MONETARY BENEFITS
▪ Access fees ▪ Royalties ▪ Licence fees ▪ Joint ownership of intellectual property rights	▪ Sharing of information ▪ Research collaboration, joint ventures ▪ Technology transfer agreements ▪ Human resources development ▪ Provision of health care ▪ Development of information infrastructures ▪ Social recognition

Sharing arrangements will depend heavily on the parties involved and the social, cultural and political particularities of the situation. Governmental, non-governmental or academic institutions and indigenous and local communities may be included in sharing arrangements. The situation in both Iceland and Tonga highlight the reality that benefit-sharing arrangements should be carefully constructed to adequately address concerns of the subject population. Failure to do so may result in total failure of the project. Mechanisms to review the contractual terms, including monetary payments, can be addressed at the outset. Appropriate dispute resolution mechanisms should be carefully considered.

[87] Lorraine Sheremeta and Bartha Maria Knoppers, "Beyond the Rhetoric: Population Genetics and Benefit-Sharing," forthcoming.

Table 15 –
Summary of Public Opinion Data Relating to Issues of Commercialization

PUBLIC CONCERNS	
United Kingdom	▪ There is a general sense that medical databases should not be owned by commercial interests (Einseidel, p. 15, citing Human Genetics Commission, 2000). ▪ About 3 in 4 people feel that new products developed using genetic information should be publicly owned (Einseidel, p. 16, citing Human Genetics Commission, 2000).
Canada	▪ Some 7 in 10 are of the opinion that legislation designed to protect privacy of health information should be applicable to both public and private sectors (Einseidel, p. 16, citing Canadian Medical Association, 2000). ▪ Canadians have shown high support for the mapping of the human genome and increasing support for the idea of patenting (Einseidel, p. 17, citing Pollara and Earnscliffe, 1999). ▪ Half of respondents are not comfortable with the idea of patents on higher life forms (Einseidel, p. 18, citing Pollara and Earnscliffe, 1999). ▪ Principles of equality and access to new products should guide the commercial process (Einseidel, p. 18, citing Pollara and Earnscliffe, 1999). ▪ Half of respondents think there is joint ownership of the genetic information between themselves and the organization that administered the test (Pollara and Earnscliffe, *Genetic Privacy Issues*). ▪ Some 43% of respondents think the biological samples are their property alone (Pollara and Earnscliffe, *Genetic Privacy Issues*). ▪ There is a deep resistance to the idea of biobanks "selling" genetic data to others doing research, even with consent. This appears to be more a view of the role of profit in health care than it is a considered decision about the particular circumstances. For instance, few have factored in the cost of gathering and storing the data when they consider the issues (Pollara and Earnscliffe, *Genetic Privacy Issues*).
Japan	▪ Support for patenting drops as the focus shifts from new plant and animal varieties to patenting existing plant, animal and human genes (Einseidel, p. 17, citing Macer, 1992).
Sweden	▪ There are ethical concerns associated with the commercialization of genetic information (Einseidel, p. 18, citing Hoyer, 2002). ▪ While the public is comfortable with the commercialization of "information technology," they are less comfortable with the idea of commercializing "gene technology" (Einseidel, p. 18, citing Hoyer, 2002).
Tonga	▪ Autogen's proposal to establish a health database in Tonga using the population's genepool sparked outrage among human rights and church organizations (Einseidel, p. 32, citing Burton 2002).
PROFESSIONAL CONCERNS	
United Kingdom	▪ Issues around the increasing numbers of links being established between public and private sectors around genetic databases have been flagged (Einseidel, p. 36, citing Human Genetics Commission, 2000). ▪ A commercial market for human DNA and genetic data is well under way (Einseidel, p. 36, citing Martin, 2000).

Summary: Commercialization

Commercialization Generally

- The objective of population genetic research is to develop new drugs and treatments for human diseases. The private sector will inevitably be involved in the process; the accrual of intellectual property rights is inevitable.
- As part of the informed consent process, research participants must be informed about the potential future commercialization of their biological materials and of the results of research.

- It is imperative that population genetic research and the associated commercialization process proceed in a manner that is both cognizant and protective of the physician–patient and researcher–participant relationships.
- The potential exists that for-profit or not-for-profit corporations that exist separately from publicly funded related research institutions may hold biological samples and associated data. It is important, therefore, that procedures concerning bankruptcy and voluntary winding up of these entities be addressed to ensure that biological samples and sensitive medical history do not become "assets" that may potentially be distributed among creditors or shareholders.

Access to Biobank Resources

- Access to biobank resources by researchers (both public and private) must depend on appropriate scientific and ethical review of proposed research.
- Researchers from Canada and abroad should have liberal access to biobank data that has been accumulated through publicly supported initiatives.
- Publicly supported population genetic biobanks should be mandated to enable as much research as possible, including research into common and rare disorders.
- Population genetic biobanks should aim to facilitate research that will ultimately benefit both the subject population and the global community.

Benefit Sharing

- The interests of the participating population and the general population must be considered separate and apart from commercial interests. To this end the interests of the population should be represented in commercial agreements.
- Benefit-sharing arrangements should be prospectively incorporated into the overall design of biobank initiatives. The public should be consulted on the issue of benefit sharing.
- Benefit sharing should be considered as a mechanism (or rather a spectrum of mechanisms) to balance the commercial interests with those of research participants in a way that is both respectful and reflective of the relative contributions to the research endeavour.

4.6 Governance

Governance refers to "those processes by which human organizations, whether private, public or civic, steer themselves."[88] The governance issues that arise in the context of biobanking are complex and broader than those inherent in "health research involving human subjects." This is because biobanking involves the interaction of private entities in a realm that is traditionally considered "public." Issues of corporate governance and corporate ethics are therefore highly relevant. In the context of biobanking, governance issues arise in and between organizations, including public and private institutions, sponsor companies, regulatory agencies, research ethics boards, researchers, research participants and the general public. The governance of biobanks is important for many reasons, not least of which because it plays a key role in ensuring accountability and in building and maintaining public trust.

Michael Yeo argues convincingly that large-scale biobanks are more akin to business organizations than to research entities as traditionally conceived and that the existing legal and ethical framework governing research is inadequate. The norms of research and research ethics have been developed narrowly and do not adequately consider the privacy and consent challenges that arise in the context of biobanking. Numerous commentators have called for stronger privacy protection and changes to the requirements for informed consent for future research on samples and associated data contained in biobanks (Yeo, p. 16). Additionally, biobanking, even if construed strictly as a research tool, differs markedly from other types of research involving human subjects. Modifications to the current legal and ethical regime or the development of a separate regulatory regime are inevitably required. This argument is bolstered when the commercial exploitation of biobanks is brought into the fray. The pressing question is whether the currently available governance tools — the law, policy and ethical norms — can adequately address the concerns arising from biobanking: Are these tools sufficiently adaptable to the biobanking context, or do further tools — legislation, regulations, policy statements specific to biobanks — need to be developed?

Public opinion data suggest that the public understands the importance of ethical governance systems in genetic research, an area that is viewed as being central to the future of medical research. This is demonstrated by the public's recognition of the importance of a collaborative effort involving government and the scientific community in developing an

[88] The Governance of Health Research Involving Human Subjects, online: www.1cc.gc.ca/en/themes/gr/hrish/macdonald/macdonald_main.asp. Executive Summary.

appropriate governance regime (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). Generally speaking, individuals do not want to choose between genetic research and protection of privacy. Rather surprisingly, if forced to choose, slightly more people emphasize research than privacy protection (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). It appears that the more people learn about genetic research and biobanking, the more likely they will conclude that the benefits of the research outweigh the risks involved. This does not mean that there is an unconditional acceptance of the free use of genetic information. Rather, the public wants to see a demonstrable link between the substance of the initiatives and the benefits to be derived. Without this link, the default position is that of strict privacy protection. If, however, the public is shown that there is impetus to improve governance and to address concerns about potential abuses or misuses of genetic information, public trust and confidence is enhanced. These data point strongly to the importance of public consultation and education on issues surrounding large-scale population genetic research.

There exists an opportunity for Canadian biobank planners to learn from the challenges faced during the development of other large-scale biobank initiatives. The experience of the Medical Research Council (MRC) in the United Kingdom is particularly instructive and should be closely examined so Canadian planners avoid similar criticisms. If Canada does embark on a large-scale population genetic database involving children, the planners must, on an inclusive and ongoing basis, engage scientists and other stakeholders in the process. In the first instance, it is necessary to address anticipated challenges to the scientific methodology and anticipate that criticism will be directed at the nature of the development process itself. It is advisable to consider whether a peer review process might be effectively employed to ensure objectivity and independence in the project development process.

Regardless of the degree of reform that is deemed necessary, large-scale biobanking clearly requires the development of an accountable regulatory framework that incorporates the legal and ethical norms governing human subject research and the evolving ethical norms of corporate governance.

Table 14 depicts, schematically representation, an overview of the complex governance environment in which biobanks operate in Canada.

Table 16 –
Overview of the Governance Environment in Which Biobanks Operate in Canada

<table>
<tr><td colspan="4">PROFESSIONAL RESPONSIBILITY
Professional and Scientific Norms

Universal Declaration of Human Rights (UN)
Declaration of Helsinki (WMA)</td></tr>
<tr><td colspan="2">Science
Various professional codes promoting: honesty, integrity, protection of the public interest, academic freedom, data sharing, peer review (etc.).
(Canadian Information Processing Society)
(Canadian Council of Professional Engineers)
(Chemical Institute of Canada)</td><td colspan="2">Medicine
Hippocratic Oath
(Provincial Colleges of Physicians and Surgeons)
(Canadian Medical Association)
(National Specialist Societies)</td></tr>
<tr><td colspan="2">Law</td><td>Policy</td><td>Ethics</td></tr>
<tr><td>Charter of Rights and Freedoms

Federal:
▪ privacy acts (public sector, private sector)
▪ CHRA Bill of Rights

Common law:
▪ privacy/
▪ confidentiality
▪ consent
▪ fiduciary law</td><td>Provincial/ territorial statutes:
▪ privacy (public sector, private sector, health)
▪ hospitals
▪ human tissue
▪ cancer registries
▪ public health
▪ statistics
▪ provincial charters and human rights acts</td><td>Federal:
▪ Tri-Council Policy Statement
▪ funding strategies
▪ (CIHR guidelines*)

Provincial/territorial:
▪ provincial policy statements
▪ university and hospital policy statements</td><td>Canada:
▪ Tri-Council Policy Statement
▪ Statement of Principles on the Ethical Conduct of Human Genetic Research Involving Human Populations</td></tr>
<tr><td colspan="2">International Law:
▪ Convention on Human Rights and Biomedicine
▪ Convention on Biological Diversity (by analogy)</td><td></td><td>International:
▪ International Ethical Guidelines for Biomedical Research Involving Human Subjects (CIOMS)
▪ Universal Declaration on the Human Genome (UNESCO)
▪ HUGO Statement on the Ethical Conduct of Genetic Research
▪ HUGO Statement on Benefit-sharing
▪ Statement on Databases (WMA)
▪ (Emerging norms of corporate ethics)</td></tr>
<tr><td colspan="4">Biobank-specific Policies And Procedures
(Biobank-specific guidelines for research ethics boards*)</td></tr>
</table>

Table 17 –
Summary of Public Opinion Data Relevant to Governance Issues

PUBLIC CONCERNS	
United Kingdom	▪ Participants stress the importance of regulations governing collection, storage, use and disposal of samples, and preferred oversight of these processes by an independent body (Einseidel, p.8 citing Porter et al, 2000).
Canada	▪ Canadians default toward the strict protection of genetic privacy but they afford high positive value to health and medical uses of their information (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). ▪ Canadians are quite open to research uses of their genetic information (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). ▪ When it comes to regulatory and ethical governance systems, more Canadians (56%) believe that the medical and research community should play the main role in determining priorities and procedures than believe the government should (41%) (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). ▪ Canadians believe that government should set legislation, but think the research and medical community are more expert and closer to the situation on the ground. Canadians want a strongly collaborative effort involving government and the scientific/medical communities (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). ▪ Canadians support for a regime that balances the strict protection of personal genetic information with facilitating access to such information to gain important corollary benefits (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). ▪ Public support for a governance regime requires consistent linkage between the substance of initiatives and the benefits to be derived, the default position (i.e., strict protection of privacy) comes into play (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). ▪ Knowledge and discussion about genetic research and genetic privacy increases the conviction that the benefits of facilitating access to genetic information outweigh the drawbacks (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). ▪ The more people know about genetic research and biobanks, the more likely they are to consider that the benefits outweigh the risks; those who are likely to hear about the issues early and engage are more enthusiastic about the benefits of genetic research (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). ▪ Focus group research suggests that, once people are given a sense that those responsible for governing these issues are thinking about them and working to improve their governance, concerns about potential abuse or misuse abate and interest in gaining the benefits increases (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 14). ▪ An overwhelming number of Canadians (96%) see genetic research as very (67%) or somewhat (29%) central to the future of medical research (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 93). ▪ Most Canadians (73%) exhibit a willingness to allow personal genetic information to be used in medical research (the percentage rises if one's identity is stripped from the database) (Pollara and Earnscliffe, *Genetic Privacy Issues*, p. 95). ▪ Concerns exist regarding donor anonymity, records being used for research purposes and the possibility of employers and insurance companies obtaining and misusing the information (Einseidel, p. 13, citing Canadian Medical Association, 2000).

Summary: Governance

- "Biobanking" is an amorphous endeavour that cuts across numerous scientific and medical disciplines. It will require innovative governance strategies and further refinement of the governance environment.
- Conflicts and inconsistencies that exist in the governance framework must be identified and appropriately addressed.
- The management structure of biobanks must be accountable and the commercial aspects of biobanking must be transparent not to undermine public support for such endeavours.

- Professional responsibility is reflected in the ethical and professional norms of the scientific disciplines and the norms of medical practice. Biobanking will require a novel multidisciplinary conception of professional responsibility.
- Law (provincial, national and international), policy statements (federal, provincial/territorial and institutional) and numerous relevant ethical statements are relevant in the governance of biobanks and ought to be carefully considered in the development of biobank-specific policies and procedures.
- Biobank-specific policies and procedures must clearly define, among other things (Yeo, pp. 19–20):
  - the precise contents of the biobank, including specimens (e.g., tissue, blood, cells or the DNA samples derived therefrom), DNA sequence data, demographic information, health information and other linked information
  - specimen and data collection procedures, including the terms of consent granted by participants
  - the extent to which individuals can be identified from the biobank data
  - the rules and procedures by which access to biobank contents is granted to researchers
  - the safeguards in place to protect the interests of those whose information is held (including security, oversight and accountability provisions)
  - the organizational structure and governing rules of the biobank.
- Research ethics boards urgently require guidance on the legal and ethical issues associated with biobanking.
- Research ethics board approval must be obtained for all proposed research protocols prior to recruitment of participants in population genetic research:
  - they must be independent and multidisciplinary and must include members of the participating population
  - they should evaluate and monitor the constitution of the biobank, review all research projects and review requests for access
  - to perform their function, they should be aware of all contractual terms between researchers and the biobank.

- Issues of capacity, competence and independence of research ethics boards must be addressed.
- There is a need to create an independent organization that would be responsible for project oversight and surveillance.
- There is a need to establish long-term monitoring and periodic auditing of biobanks.

4.7 Summary of the Main Issues

There has been a paradigm shift in genetic research from linkage analysis to large-scale population genetic research. This leads to increasing societal importance in ensuring that the ethical, legal and social issues are addressed as the research develops. There is real cause for optimism that society will reap profound benefits from innovations stemming from this second phase of the Human Genome Project. It is expected that population genetic research will enable researchers to gain a better understanding of the gene–environment interactions implicated in complex human diseases, which in turn may enable novel drug development strategies as well as the development of novel diagnostic and predictive tests. There is a need for rigorous scientific scrutiny and for careful consideration of the relevant ethical, legal and social issues. This summary will review the main issues identified in this synthesis paper and to prioritize them, where it is possible to do so.

In developing a strategy that will best meet the needs of the Canadian public, the research community, private industry and policy makers can draw from a wealth of experience developed in other nations; Canada must proceed cautiously and with an eye to the particularities of Canadian society and the regulatory regime in which biobanking is and will be situated.

At each step, it is important for policy makers to recognize that population genetic research is made possible through the participation of individual volunteers who collectively comprise a "population." The contribution of this population to the research is essential and, arguably, the population must be recognized as a partner in the research endeavour. It is from this presumption that the following conclusions flow.

Public Education and Consultation

In the first instance, a transparent public education and consultation strategy must be developed to determine **whether** Canada should promote, as a societal objective, the development of large-scale biobank initiatives. Policy makers should not exclude the possibility that Canada should not pursue large-scale population genetic research initiatives. If, however, Canada opts to pursue population genetic research after carefully reviewing all the issues, ongoing consultative strategies must be developed to ensure the continuing relevance and acceptance of such initiatives. Consultative strategies must be transparent and the results must be incorporated into the relevant projects where it is reasonable to do so. To this end, criticism that has been levied against UK Biobank is instructive. Consultation must not been seen as a "bolt-on activity to secure support for a "politically driven project."[89] Developing an appropriate education and consultation strategy will be challenging. Careful thought must be given to the development of an appropriate bundle of strategies that will yield the best possible understanding of public and stakeholder sentiment in the Canadian context. Appropriate and sustained levels of funding must be allocated to ongoing education and consultation.

Privacy and Confidentiality

Numerous factors make the issues surrounding privacy and confidentiality particularly challenging, including rapid developments in information technology, genetic research and bioinformatics as well as the ongoing tension between the desire to do beneficial health research and the need to protect personal health information. The issue of privacy is particularly complex. Specifically, there is no coherent legal framework to appropriately address the health-related privacy issues that are relevant in the context of biobanking. A very high priority must be to tackle these issues and to gain an understanding of the interactions between existing and proposed federal and provincial privacy statutes (and other potentially relevant statutes) that are implicated in biobanking. There is a possibility that careful analysis of the legal framework may compel consideration of biobank specific legislation that could more reasonably address specific privacy issues.

At the safeguarding level, it is necessary to consider whether appropriate mechanisms exist for encryption, anonymization and sharing of data. It is also worth considering, at a very early stage of project planning, optimal strategies for data collection and storage to ensure that accord with emerging international standards.

[89] *Supra*, note 1.

Informed Consent

In Canada, individual informed consent appears to be the *de facto* norm for prospective population genetic research. It is recognized that the inability of researchers and biobank planners to predict all possible future uses of human biologic materials and associated data poses a challenge to the process of informed consent. The implementation of large-scale population genetic research requires a reassessment of the current normative framework governing informed consent.

It must also be noted that population genetic research involving minor subjects is ethically and legally fraught with uncertainty. If Canada wishes to pursue the currently proposed Lifelong Health Initiative or other similar initiatives, it must address this issue.

Commercialization

A major objective of population genetic research is the development of new drugs and treatments for human disease. It is inevitable that private industry will be involved in the process and that it will seek to accumulate intellectual property rights over innovations. The challenge is to effectively reconcile the needs of industry with the needs of the broader research community and with the populations that enable such research. Numerous concerns arise in the context of the commercialization of genetic research, not least of which is the potential adverse effect on public trust. Though the public is generally supportive of genetic research, there is a real risk that overemphasis of the commercial aspect will result in a backlash against population genetic research and the commercial products developed as a result of that research.

In developing a consultative strategy, Canada is well advised to consider the development of innovative public–private collaborations that involve a “partnership” with the subject populations of genetic research.

Benefit sharing is one such collaborative mechanism. It should not be viewed as contrary to the existing intellectual property regime nor as a mechanism to curb commercial involvement in genetic research. Rather, benefit sharing should be considered as a mechanism (or rather a spectrum of mechanisms) to balance the commercial interests with those of research participants in a way that is both respectful and reflective of the relative contributions to the research endeavour.

Governance

Governance issues for biobanks arise between organizations and institutions (both public and private), sponsor companies, regulatory agencies, research ethics boards, researchers, research participants and the general public. The governance of biobanks is critically important because of the role it plays in ensuring accountability and in building and maintaining public trust. The pressing question is whether the currently available governance tools — the legal, policy and ethical norms — can adequately address the concerns arising in the context of biobanks. Are the existing tools sufficiently adaptable, or are other tools needed, including legislation, regulations, and policy statements that are specific to biobanks?

When considering the appropriateness of the current governance framework, public concerns about commercial involvement and the potential misuse of genetic information cannot be underestimated. With the increasing trend to the collection and use of genetic information, the more likely it will be that instances of misuse will occur. To assure the integrity of population genetic research, reasonable steps must be taken to understand the current governance framework and to consider ways to strengthen the protections offered to research participants and their families.

Though there is time to prospectively plan for large-scale population genetic initiatives, the reality is that research ethics boards urgently require guidance on the legal and ethical issues associated with biobanking on a smaller scale. Institutional and regional banks are being established at an ever-increasing rate as the potential value of genetic information is being realized. The Tri-Council Policy Statement provides insufficient guidance to research ethics boards that are asked to review biobank projects.

Recruitment Strategies

Methods of recruiting participants into population genetic research must be supported on scientific, legal and ethical grounds. In addition, mechanisms for participant recruitment must be developed that are specifically referable to the Canadian health care system and to patient behaviour within that system. Recruitment strategies ought to focus on the fair distribution of the benefits and burdens of population genetic research to the entire population. This is difficult in Canada because of the vast geographical size of the nation, the diversity of the populations within Canada and the relatively small population.

A OVERVIEW OF LARGE-SCALE POPULATION GENETIC RESEARCH INITIATIVES

CHARACT-ERISTIC	ICELANDIC HEALTH SECTOR DATABASE	UK BIOBANK	ESTONIAN GENE BANK PROJECT
Data collected	▪ personal health information Genotype data ▪ genealogical data	▪ personal health information ▪ genotype data ▪ physical examination	▪ personal health information Genotype data ▪ genealogical data
Follow-up		▪ Initial follow-up period is 10 years	
Study size	▪ 270 000, including children and the deceased	▪ 500,000 adults aged 45–60 years	▪ 1 million
Interested parties	▪ Icelandic government and deCODE Genetics	▪ Wellcome Trust/Medical Research Council	▪ Eesti Geenivaramu and eGeen Inc.
Consultation	▪ extensive post facto debate (local and international)	▪ in progress, though limited to date	▪ extensive up-front consultation
Population support	▪ initial support, but confidence eroded by opposition pressure orchestrated by Icelandic scientists and physicians	▪ Biobank project was modified in light of recommendations from members of public and health care professionals ▪ oversight body arose as a result of such consultation	▪ high level of population awareness and support for the project ▪ in August 2002, 76% of population knew of project and only 2% were against it
Enabling legislation	▪ act on a health sector database ▪ act on biobanks	▪ none	▪ human genes research act ▪ personal data protection act ▪ databases act
Licensing strategy	▪ exclusive licence granted to deCODE Genetics	▪ publicly owned; non-exclusive licences to be granted to researchers (public or private)	▪ eGeen is the exclusive licensee that will finance the project for the benefit of both parties
Recruitment	▪ physicians provide encrypted information to deCODE Genetics about potential participants; preliminary genealogy analysis is performed ▪ data are decrypted and returned to physicians who then contact appropriate patients, explain the research and obtain written consent	▪ physicians in participating medical centres contact their patients, explain the research and obtain written consent from individuals who want to participate	▪ physicians contact their patients, explain the research and obtain written consent from individuals who agree to participate
Access	▪ Ministry of Health ▪ commercial subscription ▪ special access for Icelandic researchers?	▪ individuals have a legal right to access their personal data. ▪ any use of material from the study by commercial organizations is subject to approval by the Scientific Management Committee and the overseeing body and must conform to relevant ethical and legal requirements	▪ only the gene donor or that person's physician can access personalized information about the gene donor. ▪ state agencies
Consent	▪ informed consent is obtained to access an individual's health record and to collect DNA samples	▪ participants must "provide written consent to enable follow-up through NHS registers, their general	▪ written informed consent is given by participants to have tissue sample, health information and genealogy

CHARACT-ERISTIC	ICELANDIC HEALTH SECTOR DATABASE	UK BIOBANK	ESTONIAN GENE BANK PROJECT
	▪ act on biobanks requires that biological samples be obtained for clearly defined purposes but gives board of the biobank the power to authorize use for purposes other than those for which samples were originally collected	practice and other medical records, for permission to use their data and blood samples for various analyses and specified and unspecified biochemical and genetic tests and for permission to recontact"	entered in the Gene Bank in a coded form ▪ specified uses include genetic research, public health research and statistical purposes that are in conformity with the law ▪ minor can be a gene donor if guardian is informed. ▪ consent may be withdrawn at any point up until sample is coded
Right of withdrawal	▪ donor may request destruction of his or her biological sample at any time ▪ data obtained and included in analysis prior to destruction of sample will not be destroyed	▪ not stated in draft protocol dated February 14, 2002	▪ participants can request destruction of all data that can be decoded
Feedback of results	▪ research findings are published on the deCODE Genetics Web site	▪ participants receive feedback on measures taken during physical examination ▪ information about progress of research will be available to all participants through newsletters, Web-based media and peer-reviewed publications ▪	▪
Oversight and surveillance	▪ Data Protection Commission, National Bioethics Committee ▪ dedicated bodies: monitoring committee; interdisciplinary ethics committee ▪ Monitoring Committee is statutorily mandated to ensure that the HSD is operated in accordance with the law	▪ independent oversight body, including lay membership, to oversee the workings of UK Biobank to conduct research activities and perform audits to ensure that data collected are used responsibly and within terms of consent obtained from participants	▪ Estonian Genome Project Foundation is chief processor ▪ chief processor must enter contract with authorized processors (eGeen) or gene researchers by which terms are set for storage, security measures, procedures for copying, distributing or destroying samples ▪ separate supervisory board and scientific advisory board ▪
Ethics approval	▪ all research protocols must be submitted to the National Bioethics Committee; it must monitor progress of the research and may stop research that goes beyond bounds of the protocol or is otherwise unethical ▪ Interdisciplinary Ethics Committee has the power to monitor ongoing research and to stop research that is not conducted in an appropriate manner	▪ not referenced in draft protocol dated February 14, 2002	▪ science committee to advise on matters of scientific validity of research carried out with gene bank ▪ consultative ethics committee oversees processing procedures of gene bank
Benefit sharing	▪ deCODE Genetics to provide Iceland a share of annual profits obtained from running database; profits to be used to promote health	▪ return of research result to biobank in exchange for use of samples ▪	▪ Estonian Genome Project Foundation holds a stake in eGeen Inc.

CHARACT-ERISTIC	ICELANDIC HEALTH SECTOR DATABASE	UK BIOBANK	ESTONIAN GENE BANK PROJECT
	services, research and development ▪ Icelandic medical records facilities to computerized at deCODE's expense. ▪ Icelandic government has full access to HSD ▪ pursuant to sub-license with Hoffman-LaRoche, Icelanders will receive free drugs that are developed using HSD for patent term		
Current status	▪ ongoing	▪ interim advisory group established; meets regularly and advises MRC on approaches to the project ▪ pilot studies to be conducted prior to commencement of main study	▪ pilot study completed in October 2002 ▪ main project commenced in spring 2003
URL	www.decodegenetics.com	www.ukbiobank.ac.uk	www.geenivaramu.ee

B EXCERPTS FROM THE GOVERNMENT RESPONSE TO THE WORK OF THE MEDICAL RESEARCH COUNCIL REPORT BY THE HOUSE OF COMMONS SCIENCE AND TECHNOLOGY SELECT COMMITTEE

Recommendation 30: We appreciate the difficulties in projecting the long-term running costs of Biobank at this stage but we are reassured to see that the issue is being actively considered now (para. 54).

AND

Recommendation 31: The Biobank is an exciting project and we commend the MRC's efforts to ensure that the United Kingdom is taking the lead in harvesting the fruits of the human genome. We are concerned, however, that funds were allocated to the project before the scientific questions over its value and methodology were fully addressed (Para 57). [Governance]

> **Government Response:** The Government endorses the Committee's commendation of the MRC in ensuring that the United Kingdom continues to take a lead in harvesting the fruits of the human genome research. Unanimity is rare among the scientific community for a major project. One of the reasons that the project has taken so long to bring to fruition (the first joint meeting held by the MRC and Wellcome Trust for scientists wishing to assist in development of the concept was in 1998) is that the funders have engaged scientists and other stakeholders on an inclusive basis from every stage. Now that the "hub" and "spokes" responsible for collection and analysis of samples and data have been selected, work will continue on finalization of a business plan and protocol and these will be widely disseminated.

Recommendation 32: It is not clear to us that Biobank was peer-reviewed and funded on the same basis as any other grant proposal. Our impression is that a scientific case for Biobank has been put together by the funders to support a politically driven project (para. 58). [Governance]

AND

Recommendation 33: We recommend that the MRC publish the comments of Biobank's peer reviewers anonymously to build confidence that the project is fully justified and supported by the scientific community (para. 59). [Governance]

Government Response: The idea of the Biobank was first raised by scientists and the project was peer reviewed. However it would not have been appropriate to review the project "like any other grant proposal." It is designed as a national resource for future research projects which cannot yet be specified in detail (though there will of course be peer review of these). The joint peer review procedure used by the funders (DH, Wellcome Trust and the MRC) involved predominantly international experts as it was agreed that this was the best way of ensuring objectivity and independence and avoiding conflicts of interest.

The Government accepts the integrity of the peer review operated by the funders in relation to the UK Biobank project. Reviewers' comments cannot be published as they were sought in confidence by funders in the normal way. Peer reviewers' comments are currently exempt from the *Freedom of Information Act* disclosure requirements which take effect in 2005. Any changes to the current practice on peer review would need to be discussed, agreed and implemented consistently by all the research councils and other stakeholders.

Recommendation 34: We believe that fully informed consent is an essential requirement for participation in Biobank. The MRC may have good grounds for not adopting the Human Genetics Commission's guidelines on consent for Biobank but it should state clearly what its position is and, if it disagrees with them, explain why (para. 60). [Informed consent]

Government Response: The government agrees that fully informed consent is paramount. Planning for BioBank has always been on the basis that fully informed consent is an essential requirement. The discussions on consent in the Human Genetics Commission report Inside Information are not guidelines but rather general points to consider. These are consistent with the principles for consent which the funders are developing for BioBank, and with the guidelines developed by an expert MRV Working Group on DNA collections, which was published following wide consultation in 2000. In practice therefore, the Human Genetics Commission's points will indeed be followed.

Recommendation 35: We fear that the project's long-term viability could be threatened if Biobank's funders fail to adopt a more open approach and engage not only the project's participants and stakeholders but the wider public (para. 63). [Consultation]

AND

Recommendation 36: It is our impression that the MRC's consultation for Biobank has been a bolt-on activity to secure widespread support for the project rather than a genuine attempt to build a consensus on the project's aims and methods. In a project of such sensitivity and importance, consultation must be at the heart of the process not at the periphery (para. 65). [Consultation]

> **Government Response:** The Government recognizes that consultation forms a vital part of the development of a project such as this. MRC and the other funders are committed to continued wide consultation as the project develops. The UK BioBank project is predicated on the willingness of volunteers to participate — broad public acceptance of the aims of the project is therefore an essential part of its implementation.
>
> Consultation with a wide range of stakeholders, including the public, has been a fully integrated part of the project planning over the last three years and this is one of the reasons that the project has taken so long to bring to fruition. Initiatives include:
>
> - informal consultation workshops with health professionals (GPs, nurses, etc.) across the country in 2001 and 2003
> - independent qualitative and quantitative research by (different) consultants in 2000 and 2002, reports of which have been published; the 2002 study was followed up, partly at the suggestion of some focus group participants, early in 2003 with further work with those social groups that had been underrepresented
> - an ethics consultation workshop in 2002, involving ethicists and special interest groups; the report has been published and has informed development of the current draft ethics and governance framework on which there will be further consultation in 2003
> - workshops for the wider research community wishing to contribute to the development of the project (in 2001 and 2002)

- presence at science festivals (e.g., BA, Cheltenham)
- meetings with Human Genetics Commission including a public forum in 2002
- a parliamentary briefing in 2003.

Most of these consultations have indicated broad public support for the Biobank concept. Comments on the ethics and governance structure and on the scientific protocol are being taken into account in developing the project further.

The Biobank's communication and consultation strategy over the next few months will focus in a number of different areas. External consultants have been commissioned by the funders to explore attitudes to the proposed ethics and governance framework for the project. In parallel, the funders will start to develop, with the newly appointed CEO for Biobank, a longer-term communications and consultation strategy for the project. This will include communications and consultation work to be done locally by the spokes with potential volunteers and health professionals, to underpin final development and piloting of the research protocol (a copy of the current draft has been available on the Biobank Web site since the summer of 2002). The CEO plans to appoint a Communications Director for the project later this year. Funders are also in the process of setting up a Public Panel. This is a group of approximately 50 people who have participated in previous consultation work commissioned by the funders and who have expressed an interest in some continued involvement.

Recommendation 37: The MRC appears to be taking a sensible attitude toward industrial involvement in Biobank. It must be made clear that all results will be in the public domain but we recognize that if new therapies are to arise from Biobank, industry's involvement is inevitable and necessary (para. 66). [Commercialization]

Government response: The Government welcomes this recommendation.

Recommendation 38: We agree with the Human Genetics Commission that Biobank's participants should be represented on the independent oversight body or on participants' panels at each regional centre. It is vital that participants play an active role in the management of the project (para. 69). [Governance]

Government response: The Government and the MRC agree that volunteer participation in oversight of the project would be helpful nationally and/or locally.

Details of the members of the oversight group will be made public. This will mean that although the identity of the volunteers and all data samples would be confidential, volunteers who are on the oversight group may be identifiable as volunteers. This would need to be adequately explained and consent obtained.

Recommendation 39: The Human Genetics Commission has recommended that the Government fund research into the encryption techniques to ensure data security. We support this view (para. 71). [Privacy and Governance]

Government response: Work is already under way within the NHS' national IT programme leading to the determination and establishment of reliable and robust standards that will ensure the confidentiality and security needs of NHS patient data are satisfied. These new arrangements will include appropriate means to anonymise, pseudoanonymise and encrypt patient data according to identified and agreed needs and that are suitable for patient data stored in databases or that is communicated electronically between information partners. The standards for achieving this security and confidentiality will be piloted and validated within arrangements for the NHS' national IT programme and will be available to the MRC for adoption as required.

DH is working closely with government security authorities including 'The Central Sponsor for Information Assurance' to ensure appropriateness of NHS methods and to achieve alignment with government advice where appropriate.

Recommendation 40: It is important that participants in Biobank are aware of the risk that police could obtain access to their data and samples before giving consent and before samples are taken. The funders should monitor to what extent this issue acts as a disincentive to participation (para. 72). [Privacy, Consent and Recruitment]

Government response: The Government agrees that participants are made aware of this issue. The position of the funders has been that police could not search the database but that they could not refuse specific access in the unlikely event of a court order. When the issue of police access was explored during consultation, people seemed accepting of this approach.

Recommendation 41: The MRC has a distinguished history and can claim credit for the high status of United Kingdom biomedical research. We commend it for valuable work it undertakes to maintain that reputation. Nevertheless, there is significant disquiet about the policies and performance of the MRC from individual researchers and organizations. We realize that we were unlikely to receive submissions from people with no grievances but we have concluded that those who have submitted evidence have legitimate concerns. We have found evidence of poor financial management and poor planning, with too many funds committed over long periods leading to large numbers of top-quality grant proposals being turned down. The MRC has introduced misguided strategies for its research support that have discriminated against young researchers and some disciplines. It has been guilty of inconsistent and inadequate communications which have hampered our ability to assess the MRC's performance and misled its research community. Combined, these have harmed the reputation of the organization and caused great resentment among and inconvenience to the research community it is meant to be supporting (para. 74).

Government response: The Government welcomes the Committee's attempt, in this conclusion to balance the MRC's continuing track record against the complaints the Committee has received.

The MRC has an excellent track record and has shown in its annual reports the MRC meets its mission as set out in their Royal Charter.

While we agree that some areas of financial management and planning could be improved, we believe that as a whole the MRC is able to plan and control its expenditure, their audited accounts have been approved by the NAO.

The Government does not agree that the MRC's research strategies are misguided. The MRC's long term strategies are developed by the MRC council, which has representatives from the scientific and medical communities, in consultation with a range of organizations including the MRC research boards and Government departments.

The Government has acknowledged throughout this response the need to pay greater attention to communication with the research community, and to evaluation of research policy and strategy.

Source: United Kingdom, Department of Trade and Industry, *"Government Response to 'The Work of the Medical Research Council' Report by the House of Commons Science and Technology Select Committee* (HC 132)," Cm 5834, June 2003, pp. 15–19.

C SUMMARY OF CANADIAN PRIVACY LEGISLATION

JURISDICTION	LEGISLATION (BILL) COVERS:	CITATION AND STATUS OF BILLS AS OF JULY 31, 2003
Federal	public sector	*Privacy Act*, R.S.C. 1985, c. P-21.
	private sector	*Personal Information Protection and Electronic Documents Act (PIPEDA),* S.C. 2000, c.5. Not applicable to private sector holdings or health information until January 2004
British Columbia	public	*Freedom of Information and Protection of Privacy Act*, R.S.B.C. 1996, c. 165, URLS: http://www.oipc.bc.ca/legislation/FOI-ACT.pdf
	private (bill)	*Personal Information Protection Act* – Bill 38 ▪ introduced in legislature on April 30, 2003 ▪ received second reading on May 1, 2003 ▪ if passed, will come into force on January 1, 2004 URLS: http://www.legis.gov.bc.ca/37th4th/1st_read/gov38-1.htm
	tissue	*Human Tissue Gift Act*, R.S.B.C. 1996, c. 211 URLS: http://www.qp.gov.bc.ca/statreg/stat/H/96211_01.htm
Alberta	public	*Freedom of Information and Protection of Privacy Act*, R.S.A. 2000, c. F-25 URLS: http://www.qp.gov.ab.ca/documents/Acts/F25.cfm?frm_isbn=077971558
	private (bill)	*Personal Information Protection Act* – Bill 44 ▪ introduced in legislature on May 14, 2003 ▪ if passed, will come into force on January 1, 2004 URLS: http://www.assembly.ab.ca/pro/bills/ba-bill.asp?SelectBill=044
	health	*Health Information Act*, R.S.A. 2000, c. H-5 URLS: http://www.qp.gov.ab.ca/documents/Acts/H05.cfm?frm_isbn=0779709268
	tissue	*Human Tissue Gift Act*, R.S.A. 2000, c. H-15 URLS: http://www.qp.gov.ab.ca/documents/Acts/H15.cfm?frm_isbn=0779716345
Saskatchewan	public	*The Freedom of Information and Protection of Privacy Act*, S.S. 1990-91, c. F-22.01 URLS: http://www.qp.gov.sk.ca/documents/English/Statutes/Statutes/F22-01.pdf
	health	The Health Information Protection Act, S.S. 1999, c. H-0.021 (Bill 29) ▪ received Royal Assent on May 6, 1999 ▪ not yet proclaimed into force URLS: http://www.qp.gov.sk.ca/documents/english/firstread/1999/bill-29.pdf
	tissue	*The Human Tissue Gift Act*, R.S.A. 1978, c. H-15 URLS: http://www.qp.gov.sk.ca/documents/English/Statutes/Statutes/H15.pdf
Manitoba	public	*Freedom of Information and Protection of Privacy Act*, C.C.S.M., c. F175 URLS: http://web2.gov.mb.ca/laws/statutes/ccsm/f175e.php
	health	*Personal Health Information Act*, C.C.S.M., c. P33.5 http://web2.gov.mb.ca/laws/statutes/ccsm/p033-5e.php
	tissue	*Human Tissue Act*, C.C.S.M. c. H180 URLS: http://web2.gov.mb.ca/laws/statutes/ccsm/h180e.php
Ontario	public	*Freedom of Information and Protection of Privacy Act*, R.S.O 1990, c. F.31 URLS: http://www.e-laws.gov.on.ca/DBLaws/Statutes/English/90f31_e.htm

JURISDICTION	LEGISLATION (BILL) COVERS:	CITATION AND STATUS OF BILLS AS OF JULY 31, 2003
	tissue	*Trillium Gift of Life Network Act*, R.S.O. 1990, c. H.20 URLS: http://www.e-laws.gov.on.ca:81/ISYSquery/IRLC7D0.tmp/2/doc
Quebec	public	*An Act respecting access to documents held by public bodies and the protection of personal information*, R.S.Q., c. A-2.1 URLS:http://www.cai.gouv.qc.ca/eng/biblio_en/bib_loi_en.htm
	private	*An Act respecting the protection of personal information in the private sector*, R.S.Q., c. P-39.1 URLS: http://www.cai.gouv.qc.ca/eng/biblio_en/bib_loi_en.htm
	tissue	*An Act respecting medical laboratories, organ, tissue, gamete and embryo conservation, ambulance services and the disposal of human bodies*, R.S.Q. L- 0.2 URLS: http://www.canlii.org/qc/sta/csqc/20030131/r.s.q.l-0.2/whole.html
	tort	*Civil Code of Quebec*, S.Q. 1991, c. 64 URLS: http://www.canlii.org/qc/sta/csqc/20030131/s.q.1991c.64.i/whole.html
Nova Scotia	public	*Freedom of Information and Protection of Privacy Act*, S.N.S. 1993, c. 5 URLS: http://www.gov.ns.ca/govt/foi/act.htm
	tissue	*Human Tissue Gift Act*, R.S.N.S. 1989, c. 215. URLS: http://www.gov.ns.ca/legislature/legc/
New Brunswick	public	*Protection of Personal Information Act*, S.N.B. 1998, c. P-19.1 URLS: http://www.gnb.ca/0062/acts/acts/p-19-1.htm
	tissue	*Human Tissue Act*, R.S.N.B. 1973, c. H-12 URLS: http://www.gnb.ca/0062/acts/acts/h-12.htm
Prince Edward Island	public	*Freedom of Information and Protection of Privacy Act*, S.P.E.I. 2002, c. F-15.01 URLS: http://www.gov.pe.ca/law/statutes/pdf/f-15_01.pdf
	tissue	*Human Tissue Donation Act*, S.P.E.I. c. H-12.1 URLS: http://www.gov.pe.ca/law/statutes/pdf/h-12_1.pdf
Newfoundland and Labrador	public	*Access to Information and Protection of Privacy Act*, S.N.L. 2002, c. A-1.1 1. received Royal Assent on March 14, 2002 2. not yet proclaimed into force URLS: http://www.gov.nf.ca/hoa/statutes/a01-1.htm
	tissue	*Human Tissue Act*, R.S.N.L. 1999, c. H-15 URLS: http://www.gov.nf.ca/hoa/statutes/h15.htm
Yukon	public	*Access to Information and Protection of Privacy Act*, S.Y. 1995, c. 1
	tissue	*Human Tissue Gift Act*, R.S.Y. 1986, c. 89 URLS: http://www.canlii.org/yk/sta/index.html (search for Yukon statutes from this site)
Northwest Territories	public	*Access to Information and Protection of Privacy Act*, S.N.W.T. 1994, c. 20
	tissue	*Human Tissue Act*, R.S.N.W.T. 1988, c. H-6 online: Canadian Legal Information Institute URLS: http://www.canlii.org/nt/sta/tdm.html (search for NWT statutes from this site)
Nunavut	public	*Access to Information and Protection of Privacy Act* (Nunavut), S.N.W.T. 1994, c. 20 as duplicated for Nunavut by s. 29 of the Nunavut Act
	tissue	*Human Tissue Act*, R.S.N.W.T. 1988, c. H-6 as duplicated for Nunavut by s. 29 of the *Nunavut Act* URLS: http://www.canlii.org/nu/sta/cons/index.html (search for Nunavut statutes from this site)

D INFORMED CONSENT: INFORMATION RELEVANT TO INDIVIDUAL PARTICIPANTS IN POPULATION GENETIC RESEARCH

Articles 2.4 and 10.2 of the Tri-Council Policy Statement (TCPS) on advise that research participants should be provided with the following information about the research protocol(s) in which they are asked to participate:

Article 2.4:

- information that the individual is being invited to participate in a research project;
- a comprehensible statement of the research purpose, the identity of the researcher, the expected duration and nature of participation, and a description of research procedures;
- a comprehensible description of reasonably foreseeable harms and benefits that may arise from participation, as well as the likely consequences of non-participation, particularly in research related to treatment, or where invasive methodologies are involved, or where there is a potential for physical or psychological harm;
- an assurance that prospective subjects are free not to participate, have the right to withdraw at any time without prejudice to pre-existing entitlements, and will be given continual and meaningful opportunities for deciding whether or not to continue to participate; and
- the possibility of commercialization of research findings, and the presence of any apparent or actual or potential conflict of interest on the part of researchers, their institutions or sponsors.

Commentary

The commentary in the TCPS relating to Article 2.4 contains a table listing "additional information that may be required for some projects." The following information is directly relevant to biobank projects:

- an assurance that new information will be provided to the subjects in a timely manner whenever such information is relevant to a subject's decision to continue or withdraw from participation;
- information on the appropriate resources outside the research team to contact regarding possible ethical issues in the research;
- an indication as to who will have access to information collected on the identity of subjects, descriptions of how confidentiality will be protected, and anticipated uses of data;
- the ways in which the research results will be published, and how the subjects will be informed of the results of the research.

Article 10.2:

- the type and amount of tissue to be taken, as well as the location where the tissue is to be taken;
- the manner in which tissue will be taken, the safety and invasiveness of acquisition, and the duration and conditions of preservation;
- the potential uses for the tissue, including any commercial uses;
- the safeguards to protect the individual's privacy and confidentiality;
- identifying information attached to the specific tissue, and its potential traceability; and
- how the use of the tissue could affect privacy.

E AN OVERVIEW OF INTERNATIONAL STATEMENTS RELATING TO PRIVACY AND GENETIC INFORMATION

United Nations, Universal Declaration of Human Rights, 1948	
Art. 12	No one shall be subjected to arbitrary interference with his privacy, family, home or correspondence, nor to attacks upon his honour and reputation. Everyone has the right to the protection of the law against such interference or attacks.
WMA, Declaration of Helsinki, 1964	
Art. 2	It is the duty of the physician in medical research to protect the life, health, privacy and dignity of the human subject.
Art. 21	Every precaution should be taken to respect the privacy of the [research] subject, the confidentiality of the patient's information and to minimize the impact of the study on the subject's physical and mental integrity and on the personality of the subject.
HUGO Statement on the Principled Conduct of Genetic Research, 1996	
	The HUGO–Ethical, Legal and Social Issues Committee recommends "recognition of privacy and protection against unauthorized access be ensured by the confidentiality of the genetic information. Coding of such information, procedures for controlled access, and policies for the transfer and conservation of samples and information should be developed and put into place before sampling. Special consideration should be given to the actual or potential interests of family members."
Council of Europe, Convention on Human Rights and Biomedicine, 1997	
Art. 10(1)	Everyone has the right to respect for private life in relation to information about his or her health.
Art. 10(2)	Everyone is entitled to know any information collected about his or her health. However, the wishes of individuals not to be so informed shall be observed.
UNESCO, Universal Declaration on the Human Genome, 1997	
Art. 5(c)	The right of each individual to decide whether or not to be informed of the results of genetic examination and the resulting consequences should be respected.
Art. 7	Genetic data associated with an identifiable person and stored or processed for the purposes of research or any other purpose must be held confidential in the conditions set by law.
CIOMS, International Ethical Guidelines for Biomedical Research Involving Human Subjects, 2002	
Guideline 18	The investigator must establish secure safeguards of the confidentiality of subjects' research data. Subjects should be told the limits, legal or other, to the investigator's ability to safeguard confidentiality and the possible breaches of confidentiality.
World Medical Association, Declaration on Ethical Considerations Regarding Health Databases, 2002	
s. 1	The right to privacy entitles people to exercise control over the use and disclosure of information about them as individuals. The privacy of a patient's personal health information is secured by the physician's duty of confidentiality.
RMGA, Statement of Principles on the Ethical Conduct of Human Genetic Research Involving Human Populations, 2003	
Art. 4	Mutual confidence between the researcher and the population is essential to reciprocity. To respect this confidence, the researchers should ensure the security and the confidentiality of the population data.

CONCLUSIONS

Eugene Oscapella concludes that the "key to benefiting from genetic information while avoiding its drawbacks lies in controlling use of information beyond the health care of the individual to whom the information relates."[3] He suggests that the ultimate protection may often lie in strictly limiting the initial collection of personal genetic information. That is valid insofar as future research is concerned. However, the genetic genie is already out of the bottle. And, as these authors have found, it is highly doubtful that existing protections against discrimination and intrusions into our individual privacy are adequate.

Biobanking is already providing the foundation for large-scale population genetic research that holds much promise, in Canada and abroad. Yet, as Trudo Lemmens and Lisa Austin have pointed out, unlike other health information, genetic information is inherently linked to a particular individual. With the assistance of computer technology, there is always a possibility that anonymous genetic information may be linked with a particular person. Indeed, their paper points out that identification is the purpose of the world's largest collection of DNA samples, which is held by the U.S. military.

The questions, therefore, are several: Do we have the appropriate safeguards in place to protect individuals from infringement on their rights? Can we preserve the confidentiality of information contained in medical records or collected in biobanks? How can we guide the storage, dissemination and commercialization of the genetic profiles of Canadians to maximize the potential health benefits for all Canadians, while at the same time protecting their privacy and human rights?

Each of these papers has also challenged us to step back and examine some of the larger questions related to biobanking in Canada: What types of genetic research and discrimination are acceptable and which are not? How far are we prepared to allow behavioural research and genetic profiling to go? What is ethical and what is not? Who decides? What checks and balances need to be put into place to govern biobanks and who will audit current and future practices?

[3] Oscapella, p. 10

Statements drawn from each of the papers collected here underscore the urgency of these questions for all Canadians:

> Inattention to or deliberate neglect of privacy and discrimination issues can turn genetics from among the most promising advances in science into a powerful weapon for undermining human rights.[4]

> Behavioural genetics research linking intelligence, criminality, attention deficit hyperactivity disorders (ADHD), and other behavioural traits to specific genes that may be more prevalent in some ethnic communities than in others creates even greater risks for stigmatization and discrimination.[5]

> A transparent public education and consultation strategy must be developed to determine *if* Canada should promote, as a societal objective, the development of large-scale biobank initiatives.[6]

While the papers collected in this volume have raised serious doubts about the protections we share, each of the authors is convinced – as are Canadians at large – that we will benefit profoundly from genetic research that is facilitated by biobanking. As a society, we must ensure that the conduct of biobanking is beyond ethical reproach. It is also incumbent upon legislators to ensure that the dividends of genetic research are shared appropriately. Canada must proceed cautiously and with an eye to the particularities of our society and the regulatory regime in which biobanking is and will be situated.

[4] Oscapella, p. 35
[5] Trudo Lemmens and Lisa Austin, "Of Volume, Depth and Speed: The Challenges of Genetic Information" (2001), p.21
[6] Sheremeta, p. 64