



Health Policy Research

Bulletin

The Next Frontier: Health Policy and the Human Genome

With the beginning of the new millennium came the news that a large part of the human genome had been sequenced. Although the knowledge that heredity plays a role in determining “health” and “illness” is not new, until now the belief was that little could be learned beyond what came from family medical histories. That is about to change.

Much work remains to understand fully the molecular processes by which heredity influences disease. However, uncovering these pathways will have broad consequences for the pursuit of health and the practice of medicine. The implications for diagnostics, preventive medicine and, eventually, therapeutics are far reaching. For example:

- Genetic prediction of an individual’s overall susceptibility to major diseases (i.e., the creation of a genetic profile) will become part of the medical mainstream in the foreseeable future. Moreover, the development of preventive and therapeutic strategies will likely follow.
- Increased genetic testing will lead to more demands for regulatory and quality control mechanisms and for enhanced health insurance coverage.
- There will be new opportunities for studying how biology interacts with the natural and social environments to trigger disease processes.

In this Issue

Genetics and Genomics

Key Policy Issues 4

An Industry Profile 13

Promises and Concerns 15

Legal Issues 18

New and Noteworthy 24

Mark Your Calendar 26

As a result, public and private sector health organizations will face an increasing number of complex challenges and opportunities. This issue of Health Canada's *Health Policy Research Bulletin* examines the policy issues associated with these new genetic frontiers, with a particular focus on the implications of genetic testing for late onset disease and investments in new resources and technologies. 🍌

Some Commonly Used Terms

As in any field of study, genetics and genomics has its own "language." Here is a sample of some of the most commonly used terms.

Biotechnology — the process of making products using living organisms or the components of living organisms — in contrast to purely chemical processes.

Bioinformatics — the application of computer and statistical techniques to the analysis and management of biological data, in particular, to complex genomic data.

DNA (deoxyribonucleic acid) — the biochemical unit of heredity and the constituent material in all genes.

Genes — the physical and functional units of heredity. They are composed of DNA sequences and are located on cellular structures known as chromosomes.

Genetics — the study of heredity and the variation of inherited characteristics.

Genetic testing — medical testing using a sample of an individual's blood or other tissue to identify specific genetic markers.

Gene therapy — the process of inserting new genetic material into an organism for the purpose of treating or controlling a genetic disease.

Genome — all of an organism's genetic material, including chromosomes, genes and DNA.

Genomics — the study of the structure and function of the genome.

Late onset diseases — gene-based diseases whose symptoms typically appear in adulthood.

Mutations — changes or alterations within a gene that may or may not be harmful.

Proteomics — the study of the complete set of proteins (the proteome) encoded in genetic material (DNA).

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

Health Canada

About the Health Policy Research Bulletin

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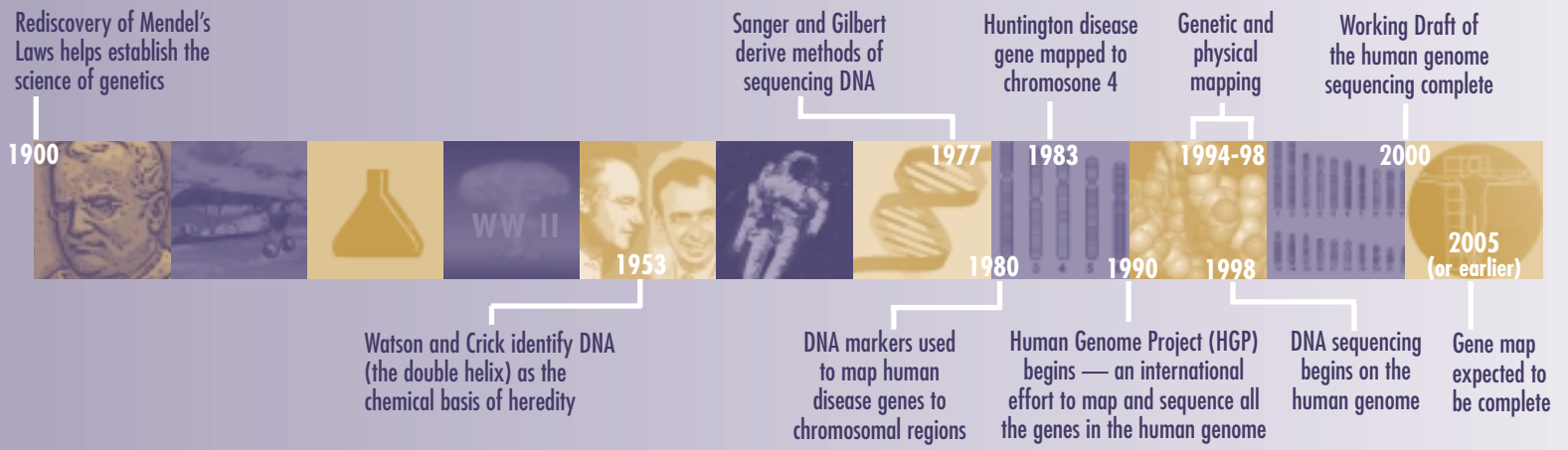
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A Century at a Glance



What Have We Learned?

The **Human Genome Project** is transforming how we see ourselves and the world around us. Here are just a few of the things we've learned so far.

We're more alike than we thought.

- The human genome has approximately 30,000 genes; the genome of the fruit fly has 13,000 genes.
- There is a great deal of genetic similarity between species — for example, 98.5 percent of the human genome is the same as that of the chimpanzee.
- Almost all (99.9 percent) of the DNA sequence is identical in every human being.
- The degree of genetic variation is almost the same between races as it is within a race.

What about the differences?

- It's not the number of genes, but the regulation of gene expression (i.e., which proteins are produced under what circumstances) that determines individual differences.
- The human genome has the capacity to encode perhaps 300,000 different proteins.

Genes are involved in some way in nearly all diseases.

- We can inherit a genetic mutation from our parents.
- We can acquire a genetic mutation during our lifetime that has not been inherited.
- We can have a genetic makeup that predisposes us to certain diseases.

What is the genetic basis of mortality?

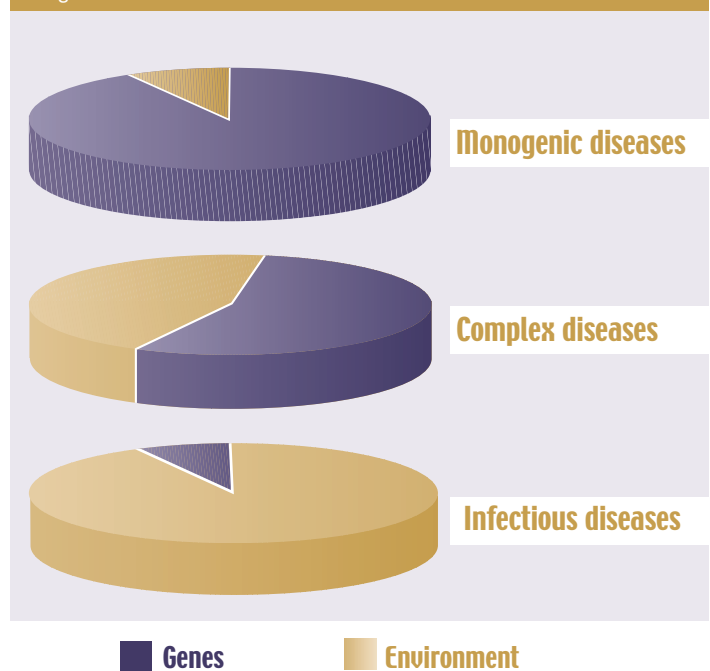
- Chromosome abnormalities account for 0.4 percent of all deaths.
- Single gene disorders account for 2-3 percent of all deaths.

- Somatic mutations (i.e., mutations acquired in one's lifetime) account for 24 percent of all deaths.
- Multi-factorial causes account for 65 percent of all deaths.

To what extent do genetics and the environment influence disease?

- As illustrated in Figure 1, the relative contribution of genes and the environment varies depending on the type of disease. 🌐

Figure 1: Influence of Genes and the Environment



Based on a presentation by Dr. Stephen Scherer, Centre for Applied Genomics, Toronto Hospital for Sick Children, and University of Toronto, given at a Health Canada symposium in March 2001.

Genetics & Genomics

Key Policy Issues

The following article is based on an interview with Phyllis Colvin, conducted by Nancy Hamilton, Managing Editor of the Health Policy Research Bulletin. Ms. Colvin is the Director of the Policy Division of the Policy, Planning and Priorities Directorate, Health Policy and Communications Branch, Health Canada. As part of her work, Ms. Colvin coordinates and sponsors the activities of the Expert Working Group on Genetic Testing for Late Onset Diseases.

Q Why is this area significant for health policy research and development?

Genetics and, more generally, biology have always been fundamental to health analysis. The *Lalonde Report* of 1974 identified biology as one of four health fields that underlie the nature and evolution of health status. Every major analysis of health since then has focussed on the role of biology.

Health Canada places genetics and biology on a list of about a dozen “health determinants,” such as income, education, environment and health care, that are known to contribute significantly to health status. Exploring genetics as a determinant of health is one of the major issues that will continue to drive health analysis.

Policy interest in genetics and biology as a potential source of health has accelerated enormously since the global Human Genome Project was established in 1990. The mapping of the human genome holds out the promise of more precise knowledge about the

linkages between genetic endowment and health status and, ultimately, the ability to manipulate biology to improve health outcomes. As the following articles demonstrate, however, the relationship between knowledge about the human genome and positive health outcomes is not necessarily direct and immediate.

Q What are some of the issues associated with genetic testing?

Even before genomics, genetic testing had considerable profile in the field of medicine. However, there is a great deal of public confusion about testing. In particular, people are confused between tests that can predict a disease long before symptoms appear and those that, at best, can identify people who may be predisposed or susceptible to a given disease, should other risk factors be present. In the latter case, genetic predisposition may be necessary but not sufficient for the disease to occur.

To date, genetic testing has been used primarily in situations involving single genes. These situations are numerous, but the actual number of



people involved is typically low. As a result, genetic testing has been confined to relatively small sectors of the population. Recently, however, genetic tests have been developed for multifactorial genetic situations, including those for breast cancer and colon cancer. Expectations are that most of the major disease-based categories of health analysis — including heart disease, cancer, diabetes and arthritis — will soon yield to genetic testing, thus creating cohorts of the “not yet ill.” It is unclear what the short- or long-term physical and psychological implications of testing will be for individuals and groups, especially as these tests are generally not predictive at some level — for example, if an environmental trigger is required for onset of the disease — and particularly with respect to the time of onset. Analysis indicates that people distinguish between genetic information and other forms of medical information, in large part, because of family, community and intergenerational implications.

Individuals who undergo genetic tests are confronted with many uncertainties, especially how to weigh the results relative to other factors contributing to their health. Moreover, genetic tests are often conducted without adequate attention to follow-up counselling and prospects for a remedy. That being said, individuals can use the knowledge gained from genetic testing to make decisions about many aspects of their lives including, for example, reproduction, employment, savings and insurance. Whatever the uncertainties, however, genetic testing is a feature of the lives of many Canadians and is expected to expand dramatically, driven by increases in both supply and demand.

Q *Why has genetic testing for late onset diseases been selected as the focus for discussion in this area?*

There are many policy issues associated with genetic testing. Some are being addressed in proposed legislation on assisted human reproduction. Others reflect the reality that genetic tests are being used earlier in

the developmental process — in children, in prenatal medicine and, most recently, at the embryonic stage. This issue of the Bulletin focuses on genetic testing for common late onset diseases as the population impact is expected to be the greatest for these diseases in the near term.

Genetic testing for late onset diseases has both diagnostic and predictive purposes. Interest in the latter is growing because access to presymptomatic information may assist in prevention, whether it is grounded in medicine, or behavioural or environmental change. At present, genetic testing for late onset diseases is generally only available to people identified as being at high risk and should not be confused with population screening. While pressure for application of genetic testing as a population screening technology is present, it is usually not contemplated until the epidemiology of a disease is well understood, adequate screening and diagnostic tests are available and, most importantly, patients have access to appropriate treatment.

While current prospects for going beyond testing to treatment and cure are limited, major investments are being made to find remedies through the development of new genetic technologies (see page 13). As this occurs, genetic testing will likely move out of the relatively restricted area of biomedicine proper to encompass health and social issues associated with large populations and genetic profiling. Genetic testing is then expected to become even more pressing from a policy perspective, requiring concerted attention to ethics, clinical validity, clinical utility and long-term physical and psychosocial impacts so that individuals, communities and governments can become informed consumers of the new testing technologies. 🌐

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Genes, Health

and

Genetic Testing

Lori Engler-Todd

Strategic Policy Directorate, Population and Public Health Branch, Health Canada

Genes and Health

Our bodies are made up of millions of cells, such as heart cells, skin cells and so on.

Contained within each cell is our inherited genetic material — our genes. Genes are the units of heredity and come in pairs; one member of each pair is inherited from each of our parents. As a result of the Human Genome Project, we know that we have about 30,000 genes in total.

The information contained in genes provides the basis for a baby's development and, along with other factors, plays an important role in maintaining health over the course of a lifetime. The majority of common illnesses, such as diabetes, heart disease and cancer result from a complex interaction of many genes with a variety of external factors. For example, by the age of 60, less than 5 percent of the population will develop an illness related to single gene defects, while about 60 percent of the population will develop a complex common illness with a hereditary influence.

Gene Mutations are Changes in the Gene

Genes are sets of instructions that can be compared to recipes or blueprints. If a gene's "recipe" is changed as a result of a mutation, the recipe may or may not turn out, depending on how the recipe is altered. For example, adding an extra egg to a cake batter may or may not make a noticeable difference in the cake, whereas leaving out the flour certainly will. Similarly, if there is too much, not enough, or none of a gene product formed, or if the product is unusual and cannot do its job, the result may be a health effect or disease state.

Gene changes that happen in the egg or sperm cell before conception can be passed on to the next generation (*germline mutations*). However, gene changes that occur in other cells during our lifetime are not inherited (*somatic mutations*). If gene changes are not naturally repaired by the body, a disease may result. For example, most cancer happens as a result of genetic errors that occur over time, leading to cells that grow out of control. Only about 5 percent of cancers are related to an inherited gene change that can be passed down from a mother or father to their child.

Many gene changes are harmless and have no ill effect on health, while others are associated with a disease state. For example, some gene changes will certainly cause disease (as with Huntington disease), some will cause disease but it will vary from a mild to a severe form (as with myotonic

dystrophy), while others result in increased susceptibility or predisposition to disease (as with an inherited cancer syndrome).

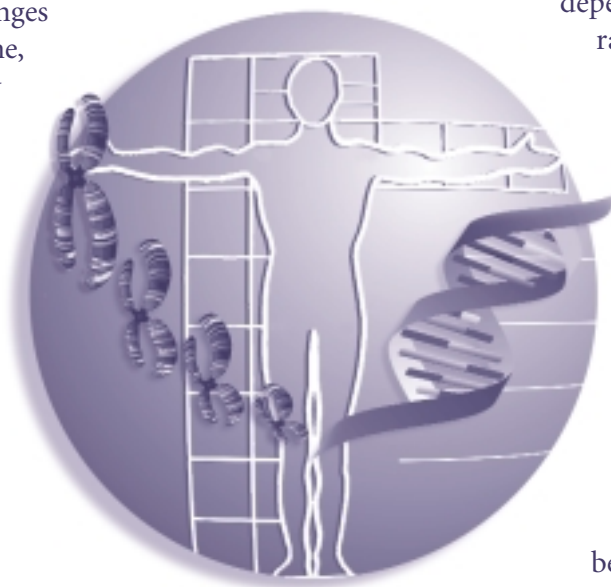
Genetic Testing for Late Onset Diseases

Late onset diseases are typically diagnosed in adulthood and do not include health problems present at birth or those that develop in childhood. Genetic testing involves taking a sample and looking for changes within a specific gene. As with the recipe analogy, you need to know how to read the recipe or what the recipe makes in order to know if it has been changed. When genetic diseases are caused by changes at the same place in one gene, testing can be simple, quick and inexpensive. For other conditions, gene changes may happen anywhere in the gene, be unique to a family, or simply be too difficult to find.

Genetic testing may be offered to confirm a diagnosis in a person who is showing symptoms. It can also be offered to a person who has not been clinically diagnosed with, nor has any symptoms of, a medical condition. This is known as *predictive* or *pre-symptomatic* genetic testing and is used to estimate the risk that a given disease will develop. Such a risk may be anywhere from as low as *the risk in the general population*, to as high as *close to 100 percent*. In some cases, the results of the test may be inconclusive.

As we are each unique, predictive genetic testing cannot tell people at what age symptoms will appear or the severity of these symptoms, if and when they do appear. Consequently, there is a potential for psychological and emotional harm from such testing, especially if there is an unrealistic expectation of

Clinical utility measures the benefits and risks of early detection in those diagnosed with a disease, as well as the overall benefit to those screened. It determines whether illness or death can be avoided by actions triggered by test results.



treatment. Meeting with a genetic counsellor beforehand can help a person arrive at a decision that they are most comfortable with, taking into account risks and benefits. Furthermore, in the absence of a cure, preventive measures or proven treatment, the clinical utility of predictive and susceptibility testing for the population and the individual needs to be examined.

Clinical Utility

Clinical utility measures the benefits and risks of early detection in those diagnosed with a disease, as well as the overall benefit to those screened. It helps determine whether illness or death can be avoided by actions triggered by test results. It is important to note that the utility of genetic testing for late onset disease varies with each disease and depends on such variables as how accurately the risk is measured and the availability of risk management strategies for both prevention and treatment, within the context of the life experience and risk perception of the individual considering the testing. The glossary in Table 1 outlines important measures in risk calculation for genetic susceptibility and clinical validity.

There is debate about the usefulness, as well as the perceived benefits and potential harm, of genetic testing for a disease that may never manifest in a person's lifetime. In the absence of genetic testing, individuals from families suggestive of an inherited illness are assigned risks based on personal and family history. Such individuals must make important decisions about having a family, possible medical or surgical interventions and lifestyle choices, not knowing if they inherited the illness. Notably, some individuals would choose to pursue information from genetic testing while others would not. ▶

Genetic Testing for Common Diseases

As discussed in the article on page 4, genetic testing in Canada is currently offered on a non-mandatory basis to individuals who are at an increased risk of single gene disorders. In the future, genetic testing may become available for common diseases with a hereditary component involving multiple genes. Such testing, which is extremely complex, may become more readily available to people with an average risk of developing a common disease. The hope is that this could lead to new targeted therapies and personalized lifestyle and environmental assessments, resulting in improved population health status. On the other hand, there is concern that private industries holding gene patents may over-promote the benefits of genetic testing which could lead to escalating health care costs. 🌀

Testing in Canada is currently offered on a non-mandatory basis to individuals who are at an increased risk of single gene disorders.

References

- Baird PA. Will genetics be used wisely? *ISUMA* 2001; 2(1) (<http://www.isuma.net>).
- Baird PA, Anderson TW, Newcombe HB, Lowry RB. Genetic disorders in children and young adults: a population study. *Am J Hum Genet* 1988; 42: 677-9.
- Bell J. The new genetics in clinical practice. *BMJ* 1998; 316: 618-20.
- Collins FS, McKusick VA. Implications of the human genome project for medical science. *JAMA* 285(5): 540-4.
- Evans PE, Skrynia C, Burke W. The complexities of predictive genetic testing. *BMJ* 2001; 322: 1052-6.
- Jones KL. *Smith's Recognizable Patterns of Human Malformation*, 5th edition. WB Saunders Company, 1997.
- Offit K. *Clinical Cancer Genetics: Risk Counselling and Management*. Wiley-Liss Inc., 1998.
- Vastag B. Experts wrestle social, ethical implications of human genome research. *JAMA* 285(6): 721-2.

Table 1: Glossary

false negatives — those with an underlying increased risk who have a negative genetic test result

false positives — those with an average risk who have a positive genetic test result

positive predictive value (PPV) — the probability an individual with a positive test result will be susceptible

reliability — quality assurance measures, including staff training, standards, etc.

sensitivity — the ability of a genetic test to identify those *with* the disease susceptibility (individuals with susceptibility who test positive)

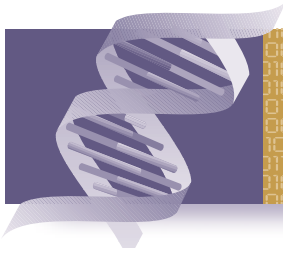
specificity — the ability of a genetic test to identify those *without* the disease susceptibility (individuals at average risk who test negative)

true negatives — those with an average risk who have a negative genetic test result

true positives — those with an underlying increased risk who have a positive genetic test result

validity — sensitivity and specificity

(Taken from Offit K. *Clinical Cancer Genetics: Risk Counselling and Management*. Wiley-Liss Inc., 1998.)



Using Canada's Health Data is a regular column of the Health Policy Research Bulletin highlighting some of the methodologies commonly used in analyzing health data. In this issue, we examine how measures of "risk" and "probability" are used.

Risk of Colon Cancer and Genetic Testing

Ruolz Ariste, Applied Research and Analysis Directorate of the Information, Analysis and Connectivity Branch, Health Canada, and Lori Engler-Todd, Strategic Policy Directorate, Population and Public Health Branch, Health Canada

Inherited Susceptibility to Colorectal Cancer

Most cancers (90 to 95 percent) are not inherited. However, based on personal and family history, some individuals appear to have a greater likelihood of developing cancer than what is seen in the general population. One example of an inherited cancer syndrome is called Hereditary Non-Polyposis Colorectal Cancer (HNPCC). HNPCC is associated with at least five different genes, including one called MSH2. Of those HNPCC families with an identifiable gene alteration, MSH2 is involved in about 50 percent of cases. When both genders are considered together, colorectal cancer ranks as the second most frequent cause of cancer deaths in Canada.

Risk and Probability

In the medical field, the term "risk" is often used to designate the likelihood of the occurrence of undesirable episodes — for example, the probability that someone is diagnosed with colorectal cancer. However, this is only one dimension of risk. A broader approach to risk goes beyond the pure probability version to one that incorporates the consequences as well. Once an event or a risk factor has been identified, two conditions must be true in order for a health risk to exist. The event must have a likelihood to occur and it must have some adverse effect on health. So, risk (R) relates to the measurement of the probability (P) that an event will occur over a specified time period (e.g., a year or a lifetime) for a specific group, combined with the severity of the damage (D) to

human health resulting from exposure to the event, or $R = P \times D$. This is the *absolute* risk.

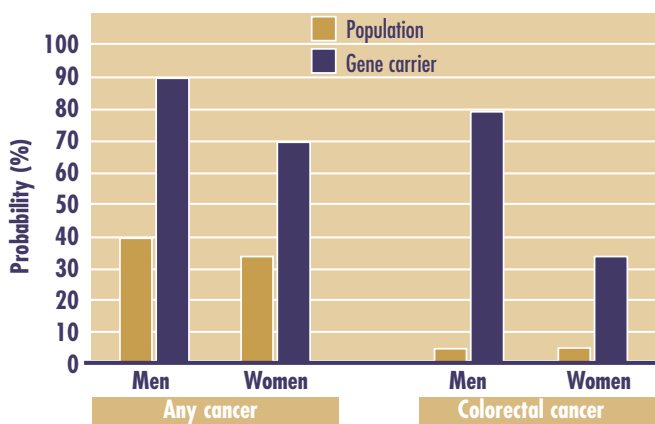
The lifetime probability of developing colorectal cancer for the general population is 6.3 percent and 5.5 percent, for men and women respectively (see Figure 1). This represents the possibility that damage (not necessarily fatal) due to colorectal cancer will occur over the course of a lifetime. Where the event is observed, the damage to health can take several forms, resulting in a range of effects from reduced quality of life to death.

Although the probability that the ultimate damage (death) will occur is often known, this is less true for reduction in quality of life. Moreover, an appropriate evaluation of the damage to health presupposes that we have a reference value for an ideal quality of life. Such a reference value is controversial, which makes it difficult to formally measure the absolute risk that any particular disease poses for human health. Without a good measure of absolute risk, comparisons across diseases are difficult if not impossible. For illustration purposes, if AD represents the anticipated average damage from colorectal cancer, the risk facing each Canadian man and woman is $0.063 \times AD$ and $0.055 \times AD$ respectively (supposing that the average damage for the two groups would be the same). The limited amount of information on average damage explains why the term "risk" is often used to mean the probability alone, as described above. For the rest of this article, the average damage will be held constant and identical for the two groups, implying that any variation in the probability leads to the same variation in the risk.

Increased Risk for HNPCC Families

Genetic testing for the diagnosis or prediction of cancer as an inherited illness is relatively new. Men and women who carry an HNPCC gene mutation in MSH2 have a probability of about 80 percent and 35 percent, respectively, of developing colorectal cancer. This means that the *increased lifetime risk* of colorectal cancer for men and women, respectively, is approximately 74 percent and 30 percent (the difference between a gene mutation carrier and the general population). ▶

Figure 1: **Risk of Developing Colorectal and Other Cancers for the General Population and for MSH2 Gene Mutation Carriers**



In the Canadian general population, the lifetime probability of developing *any type of cancer* is about 40 percent for men and 35 percent for women. The lifetime probability to develop *any cancer* for an individual with HNPCC is about 91 percent for men and 69 percent for women and may happen at a younger age than in the general population. Thus, the *increased lifetime risk* of any cancer for an MSH2 gene mutation carrier is about 51 percent for a man and 34 percent for a woman.

Figure 1 presents the probabilities of developing colorectal and any cancer for the general population and for MSH2 gene mutation carriers.

Colorectal Cancer Test Prediction and Interpretation

Increased risk is closely associated with the term *relative risk* since they both involve two groups of people. Relative risk is a statistical comparison between two groups. It is the ratio of the absolute risk of the exposed group to that of the unexposed group. Relative risk is used to determine if a specific risk factor or disease is associated with an increase, decrease or no change in the disease rate in those groups. Using the example of an MSH2 gene carrier female whose lifetime absolute risk of getting any cancer is 69 percent and any female in the Canadian population whose lifetime absolute risk of getting any cancer is 35 percent, the relative risk of the MSH2 gene carrier female is 1.97 (0.69/0.35). This means that her likelihood of

developing cancer is about *two times higher* than a woman in the general population. However, the use of relative risk can be misleading since a small absolute risk for the unexposed group may result in a large multiple of that risk for the exposed group, even if the absolute risk for that exposed group remains very small.

As noted in the example above, a woman with an MSH2 gene mutation has about a 69 percent risk of developing any type of cancer during her lifetime. In the general population, more people survive a diagnosis of colorectal cancer than die as a result of it. Provided that MSH2 gene mutation carriers have comparable survival rates to the general population, there remains a greater likelihood of dying *from all other causes of death combined* if you add up all other threats to life. Risk management strategies such as eating more leafy green vegetables, less fat and red meat, smoking cessation, and cancer surveillance (leading to early detection) all contribute to reducing the risk of developing, and of dying from, colorectal cancer. However, other predictive genetic tests may have no proven risk management strategies.

Genetic testing is subject to two types of errors: *false positive* and *false negative* (see page 8). Those errors are both related to the chosen level of significance of the test which is the probability that the test indicates erroneously the presence of a disease. There is a *tradeoff* between those two errors. The more a lab technician protects him or herself against false positive errors by choosing a low level of significance, the greater the chance of false negative error. 🌟

References

- Aubert B, Lasserre P, Ariste R. Cadre d'analyse : Les dimensions du risque. Dans : *Le projet de gestion intégrée des risques au CIRANO*. Centre interuniversitaire de recherche en analyse des organisations, Décembre 1999.
- Canadian Cancer Statistics, 2001 (<http://www.cancer.ca>).
- Froggatt B, Green J, Brassett C, Evans DG, Bishop DT, Kolodner R, Maher ER. A common MSH2 mutation in English and North American HNPCC families. *J Med Genet* February 1999; 36(2): 97-102.
- Gilpin C, Children's Hospital of Eastern Ontario. Personal communication.
- Health Canada. Health Canada Decision-Making Framework for Identifying, Assessing, and Managing Health Risks. August 1, 2000.

A Population and Public Health

Perspective

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Introduction

Of the developments arising from the “gene revolution,” genetic testing has been one of the most readily applied within clinical practice. Only now, however, are we “getting a handle” on the extent of genetic testing in Canada and the implications of testing for patient management, the provision of health services and the promotion of health and prevention of disease. This article presents the results of a recent survey of genetic laboratories in Canada and discusses the implications of genetic testing for late onset diseases from a population and public health perspective.

Survey of Genetic Laboratories

Health Canada’s Centre for Disease Prevention and Control recently conducted a survey of genetic laboratories to assess the extent of genetic testing for late onset diseases in Canada and to obtain information about the laboratories providing this type of testing. Most laboratories were hospital-based, while some were in universities and research centres. The few commercial laboratories that were identified as possibly conducting genetic tests for late onset diseases did not reply to the survey.

The laboratories reported 50 different genetic tests for late onset diseases. Over 18,000 of these tests were performed in 1999 (an underestimate as only 72 percent of identified labs responded). The most commonly reported tests were for gene variants associated with thrombophilia (a tendency for blood clots), hereditary hemochromatosis (a disorder that results in excessive accumulation of iron in the body), and breast and ovarian cancer.

Preliminary data indicate that genetic testing for late onset diseases is increasing in Canada. More tests are in the development stage and will be offered within the next five years, especially for cancer, diseases of the circulatory system and degenerative diseases of the nervous system.

Implications for Population and Public Health Activities

Surveillance and Risk Assessment

As more links between genetic factors and disease are identified and as clinicians increasingly make genetic tests a routine part of their practice, the need for surveillance and risk assessment activities increases. Surveillance helps to determine the population frequency of genetic variants that predispose the population to specific diseases as well as the amount of illness that is attributable to genetic factors.

Risk assessment activities examine the contribution of genetic risk factors to disease outcomes relative to other infectious, chemical, physical, social and lifestyle factors. This information is crucial to prevention and intervention efforts. A good example is the identification of the significantly increased risk of venous thrombosis in women using oral contraceptives who have mutations in the prothrombin gene or in the factor V Leiden gene. Genetic testing can provide useful information for counseling women who develop thrombosis about future methods of contraception. ▶

Evaluation

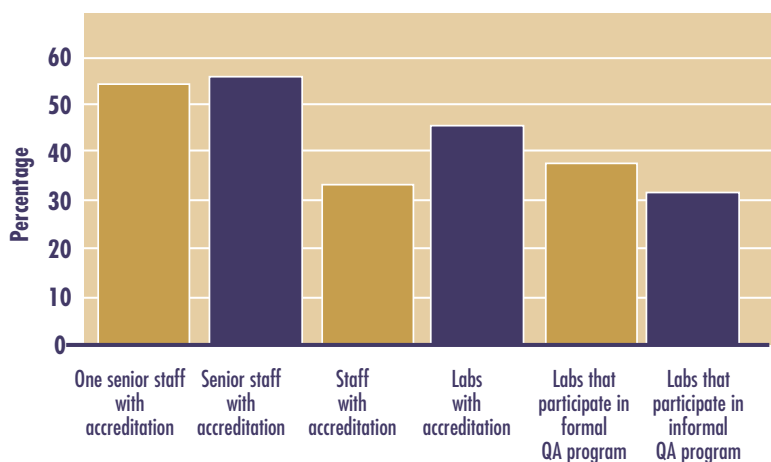
Two issues need to be addressed with respect to evaluation. First, the clinical validity and utility of genetic tests (see page 7) need to be assessed to determine a test's accuracy, safety and effectiveness. As more tests become available in clinical settings, further research is needed to evaluate the impact of using genetic tests and services with different populations.

Second, the efficacy of follow-up interventions — be they preventive, surgical, pharmaceutical or other — that are undertaken after a person is identified as being at increased risk will need to be assessed. For example, the long-term effects of a prophylactic mastectomy to reduce breast cancer risk must be weighed against the effects of living with the knowledge of being at increased risk of breast cancer. Such assessments will become increasingly important as more options are presented to the high risk sub-populations identified with genetic tests and/or as commercially distributed tests become more widely available.

Quality Management

Quality assurance standards for genetic testing must be developed. A survey of Canadian genetic laboratories showed that there is substantial variability across Canada in laboratories' participation in quality assurance and accreditation programs (see Figure 1). Participation in these programs is voluntary in Canada, except in Ontario, where laboratories must participate in programs offered by the Quality Management Program — Laboratory Services (formerly Laboratory Proficiency Testing Program). Health

Figure 1: **Participation in Accreditation and Quality Assurance Programs**



Canada's Centre for Chronic Disease Prevention and Control is currently developing a Genetic Testing Quality Management System to monitor genetic testing in Canada.

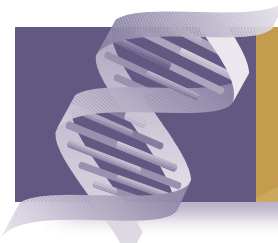
Regulation related to genetic testing will also have to be considered. Traditionally, genetic tests have been classified as “home brew” tests, meaning that laboratories prepare their own version of the test. These tests do not currently fall within the jurisdiction of the Medical Devices Regulations (MDR). Only when a genetic test is developed for commercial distribution and sale in Canada does it fall under the *Food and Drug Act* (FDA) or the MDR. To date, no applications have been received for the licensing of medical devices for conducting genetic tests under these regulatory platforms.

Communication and Dissemination

Finally, communication and dissemination of information related to genetic testing for late onset diseases is essential for educating both the public and health professionals. Professionals will play an important role in interpreting information related to genetic testing for late onset diseases, disease prevention and health promotion, especially testing that introduces complex concepts of risk, lifetime probability and potential preventive measures to reduce risk. 🌐

References

- Carballo M. Scientific Evaluator. Medical Devices Bureau, Therapeutic Products Programme, Health Products and Food Branch, Health Canada. Personal communication, May 2001.
- Fink L, Collins F. The Human Genome Project: Evolving Status and Emerging Opportunities for Disease Prevention. In: Khoury MJ, Burke W, Thomson EJ (Eds.). *Genetics and Public Health in the 21st Century: Using Genetic Information to Improve Health and Prevent Disease*. New York: Oxford University Press, 2000: 45-59.
- Holtzman NA, Watson MS (Eds.). *Promoting safe and effective genetic testing in the United States: final report of the Task Force on Genetic Testing, 1997* (http://www.nhgri.nih.gov/ELSI/TFGT_final/).
- Martinelli I, Sacchi E, Landi G, Taiolo E, Duca F, Mannucci PM. High Risk of Cerebral-Vein Thrombosis in Carriers of a Prothrombin-Gene Mutation and in Users of Oral Contraceptives. *N Engl J Med* 1998; 338: 1793-7.
- Rivard GE. General Counsel, Legal Services, Health Canada. Personal communication, May 2001.
- Vandenbroucke JP, Koster T, Briët E, Reitsma PH, Bertina RM, Rosendaal FR. Increased risk of venous thrombosis in oral-contraceptive users who are carriers of factor V Leiden mutation. *Lancet* 1994; 344: 1453-7.



Did You Know?

Did You Know is a regular column of the Health Policy Research Bulletin examining aspects of health research and data that may be subject to misconceptions. In this issue, we examine the state of development of gene-based technologies.

New Resources and Technologies: An Industry Profile

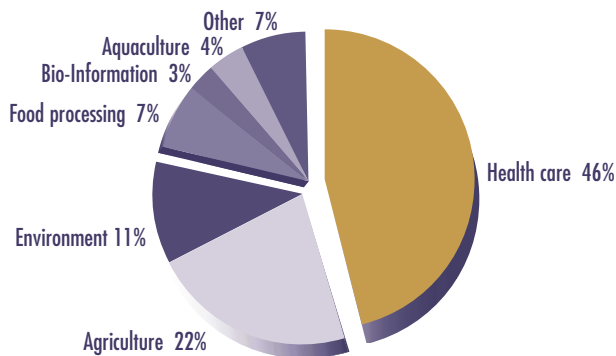
Michael Silverman, Policy, Planning and Priorities Directorate, Health Policy and Communications Branch, Health Canada

The sequencing of the human genome has generated considerable excitement about the potential for developing new gene-based therapies. In fact, many people believe that these therapies are “just around the corner.” A study of the Canadian biopharmaceutical industry conducted in 2001 by BioteCanada on behalf of Health Canada helps to shed some light on the new technologies.

Company Distribution

It is true that significant resources are being invested in biotechnology (e.g., \$594 million in 1997-98). According to a 1998 survey conducted by Statistics Canada, the Canadian biotechnology industry consists of a core of 282 firms, 25 percent of which are publicly traded. It is interesting to note that the greatest concentration of these firms (nearly 50 percent) is in the health care sector (see Figure 1).

Figure 1: Company Distribution by Sector



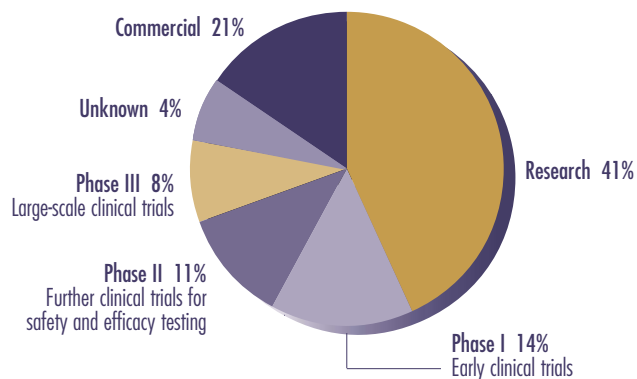
Source: Statistics Canada, The Biotechnology Firm Survey, August 1998.

The Production Pipeline

Historically, research has concentrated on the development of therapeutics and vaccines as a way of treating various diseases. However, genomics and proteomics research are closing in on and, in some cases, eclipsing

traditional areas of research. Despite this rapid increase, however, research in genomics and proteomics has yet to lead to the widespread availability of new gene-based therapies. As Figure 2 shows, the majority of products in the biopharmaceutical sector (two-thirds) are still in the early stages of development, that is, Phase II or earlier.

Figure 2: Biopharmaceutical Stage of Development (Totals)



It should be pointed out, however, that products are at very different points within the production pipeline. For example, over 70 percent of products under development in the treatment of cancer are in the early stages of research, while a substantial percentage of diabetes products are in the later stages of development.

Becoming Commercially Available

Finally, as an increasing number of gene-based therapies become commercially available, it will be important to consider the application of patents to these new types of health products. Canada's *Patent Act* allows for the patenting of genetic tests and other gene-based products. Patents cannot be granted for substances that occur in nature, but can be granted for substances derived from nature. Thus, a gene can only be patented if it has been isolated from its natural source, been purified and shown to have a specific utility. In this way, the *Patent Act* seeks to balance patent protection while creating conditions that will allow science and business to realize the promise of improved medical treatment.

References

BioteCanada. *Domestic Biopharmaceutical Foresight: A Forecast for the Biotechnology Health Care Industry in Canada*. Commissioned by the Policy Division, Health Policy and Communications Branch, Health Canada, March 2001.



Genetic Testing for Late Onset Diseases

Following are excerpts from an interview with Justice Jean-Louis Baudouin, Chair of the Expert Working Group on Genetic Testing for Late Onset Diseases. Justice Baudouin, a judge of the Quebec Court of Appeals, has been influential in the development of policies bridging the fields of medicine and law, formerly serving as Chair of the federal Discussion Group on Embryo Research. This group laid much of the groundwork for Health Canada's policy on assisted human reproduction.

Q *Can you tell us about the mandate of the Expert Working Group and how it is constituted?*

The mandate of the Working Group on Genetic Testing is two-fold: to review the current status of genetic testing in Canada and, perhaps more importantly, to provide Health Canada with a general survey of the medical, legal, ethical, social, psychological and cultural issues that genetic testing raises. Truly multi-disciplinary, the group includes physicians, scientists, lawyers, ethicists and others with an interest in the issue.

Q *What issues should be considered in determining whether Canada's health system should accept, promote and incorporate genetic testing for late onset diseases?*

Many issues must be considered, some purely technical. Most of them, however, concern how genetic testing for late onset diseases can be made available to the Canadian population and under what precise conditions. Of course, the adequacy and quality of these tests is crucial, as is their widespread availability. With the recent progress and refinements in

testing, the scientific community has raised a number of new issues that must be taken into consideration. One of the Committee's concerns is to prevent these tests from being used to gather information for non-medical purposes and to indirectly promote positive or negative eugenics.

Q *What are the key policy issues related to genetic testing?*

Many of the policy issues are directly related to their ethical and legal contexts. For example, the issue of whether or not tests could be required by employers and insurance companies is already under close scrutiny in several European countries. Canada needs to explore this issue as well. Preserving confidentiality is also a fundamental concern.

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Q *What research has the Expert Working Group undertaken and how will the results be used to address key policy issues?*

The Committee's work has been helped considerably by a number of important research papers prepared by experts in the ethical, legal, scientific and policy fields. This research will most likely be made available when the Committee's report is finalized within the year. 🌀

Promises and Concerns

Assistant Professor, Concordia University, an ethicist teaching courses in social and applied ethics. This article is based on a document entitled "Genetic Testing for Late Onset Diseases: Current Research Practices and Analysis of Policy Development." Prepared as background for the May 2000 meeting of the Expert Working Group on Genetic Testing for Late Onset Diseases, the paper draws on European and North American literature.

Genetic testing for late onset diseases promises to predict the onset of disease many years before it occurs. This type of testing requires careful thought and analysis at many levels as it raises concerns about health issues, as well as a broad range of social and ethical dilemmas. Among the questions to be considered are:

- Is genetic testing for late onset diseases a service that is ready to contribute to human health and well-being?
- Should the Canadian health care system accept and promote genetic testing for late onset diseases?

This article sets out both the promises and concerns related to genetic testing for late onset diseases. It is intended to demonstrate the ambiguous nature of this technology and highlight the need for public education, a heightened understanding by policy makers of the values shaping decisions about genetic testing and regulatory measures to address specific challenges to the health care system.

The Promises

One of the most unique aspects of the union of molecular genetics and human medicine is the possibility of predicting diseases that will occur sometime in the future. Although certain diseases have long been associated with heredity, we are now beginning to understand *why* and *how* this occurs and *where* the root of that inherited disease lies. For example, the root of Huntington disease is a gene lying on "chromosome #4." The first completely dominant human genetic disease to come to light, Huntington disease is also one of the first diseases for which presymptomatic testing became available. Advances in the Human Genome Project and post-genomic research will expand the number of ▶



presymptomatic tests available, not only for the relatively rare single-gene disorders, such as Huntington disease, but also for the more common polygenic (dependent on the interaction of *numerous* genes) and multifactorial diseases, such as cancer, diabetes, atherosclerosis, cardiovascular disease and psychiatric disorders.

If treatment is not currently available, one might question the benefit of genetic testing for late onset diseases. However, discovering that one will or may develop a disease at some time in the future could motivate a person to monitor his/her health and, if possible, take preventive measures. Testing may also provide information that can be used in making decisions about marriage, having children or taking out life insurance.

The Concerns

There are also some significant medical, social and ethical concerns about genetic testing for late onset diseases. On the basis of current medical knowledge, effective methods are not available to prevent the appearance of late onset diseases or to treat them when they do appear. Also problematic are *false positives* (tests that erroneously indicate the *presence* of a genetic condition) and *false negatives* (tests that wrongly indicate the *absence* of a genetic condition).

Another potential danger is the premature integration of genetic tests into clinical practice. This may be motivated by the promise of economic returns or the public's desire for treatment options. These factors were clearly at play in the rapid uptake of genetic testing for breast cancer in the United States after the 1995 announcement that an alteration in the gene BRCA1 indicated a high risk for the disease.

Fear of Discrimination

Socially, there is concern that genetic testing for late onset diseases may lead to discrimination as people are "marked" by a disease they may or may not develop. This is due, in part, to a general lack of understanding about the potential benefits and dangers of genetic

testing. The meaning of genetic testing for late onset diseases is widely misinterpreted because concepts such as risk, probability, sensitivity, specificity and predictive value are difficult to understand, especially in the context of one's personal health.

Informed consent is also an issue. A contributing factor is the direct marketing of genetic tests to the public, which exacerbates existing problems of quality control, accuracy, confidentiality and providing genetic information without proper counselling and follow-up. Issues such as variability in genetic testing, lack of a testing standard, lack of quality control and performance testing, and a shortage of genetic counsellors increase the probability that the results of genetic tests will be misinterpreted or misunderstood.

Notable as well is the perceived link between eugenics and prenatal testing. While there are good reasons for prenatal testing, there is also a fear that prenatal genetic testing will push parents to want the "perfect" offspring, free of current and future genetic diseases. In other words, if the goal is to reduce the number of "defective" offspring, one might ask how this differs from eugenics.

Privacy and Confidentiality

Ethical issues abound with genetic testing for late onset diseases. Privacy and confidentiality concerns are directly related to the unique situation of asymptomatic persons testing positive for late onset diseases. Three specific dilemmas arise: 1) the "intergenerational"

Figure 1: Promises and Concerns of Genetic Testing

Promises	Concerns
Predicting diseases.	Treatment most often not currently available.
Understanding <i>why</i> or <i>how</i> diseases are hereditary.	Premature integration of genetic tests into clinical practice.
Getting to the <i>root</i> causes of disease is an important first step to treatment.	Biological determinism and the danger of eugenics.
Knowing about future disease is an important factor in planning one's future.	Confidentiality and the potential for new forms of discrimination.

nature of genetics, which reveals unsolicited information about offspring and family members; 2) the lack of clarity about what constitutes medical and non-medical information; and 3) third-party interest in genetic information, which raises the very real issue of genetic discrimination. Depending on where one resides, an individual and/or family member may be denied social benefits, such as education, work, and health or life insurance, based solely on his/her apparent variation from the “normal” human genotype.

Policy Implications

Unlike some medical breakthroughs that provide fairly uncontested and immediate benefits, there is some uncertainty about the implications of genetic testing for late onset diseases. It is far from settled whether the advantages of the new technology outweigh the disadvantages. This ambiguity will have a considerable impact on policy decisions.

The Need for Public Education

Whatever direction health care policy moves in relation to specific genetic tests, Canadians must be able to make *informed* decisions. Therefore, education needs to take place at two levels. First, people need to understand what is at stake in genetic testing when they are branded as “ill,” even though no symptoms have appeared. Awareness must be heightened about the psychological and social effects of knowing about a future disease when there is no treatment and

when that knowledge may have a profound impact on family members who have not chosen to undergo testing. The fact that genetic testing is easy, simple and fairly non-intrusive may promote an uncritical attitude that should be guarded against, again through education. People need to know that genetic testing is far from certain. As well, consumers need to be aware of the potential conflict of interest between their desire for certainty and the inclination for patent holders to over-promote a genetic test.

Whatever direction health care

policy moves in relation to specific

genetic tests, Canadians must be

able to make *informed* decisions.

Promoting Public Debate

A second level at which education needs to take place to promote informed consent is at the policy level. Policy makers need to understand precisely what values are shaping decisions about genetic testing. Promoting public debate will help to identify both conflicting views and the common ground from which Canadians can shape their future.

Also important is an increased understanding of Canada’s regulatory needs for genetic testing. For example, a standardized system of quality management and performance testing is required, as are uniform mechanisms to measure when and for whom a test is suitable. Furthermore, all Canadians need to have access to genetic testing when the benefits clearly outweigh the risks. 🌐





Legal Issues in

Genetic Testing for Late Onset Diseases

Guidance from Human Rights

Abstracted from a document by Derek J. Jones, independent scholar and lecturer in health law and the Bioethics Program of McGill University, commissioned by Health Canada for the Expert Working Group on Genetic Testing for Late Onset Diseases

Several human rights standards in law are relevant to genetic testing. Some date from antiquity; others are decidedly modern.

Human Dignity

Human dignity is a foundational principle of post-war public international law, as enunciated in the 1945 UN Charter and the 1948 UN Universal Declaration of Human Rights. Genetics is not mentioned in any of these documents. Still, the Universal Declaration of Human Rights helped to inspire the 1997 UNESCO General Declaration on the Human Genome and Human Rights.

While human dignity is not explicitly mentioned in the *Canadian Charter of Rights and Freedoms*, the Supreme Court has held human dignity as a touchstone value that animates human rights norms.

Privacy

The federal *Privacy Act* offers protections for the use of personal information. However, the *Act* was not designed to deal with the challenges posed by genetic testing. Similar limitations have prompted other

nations, such as the United States, Norway, Denmark and the Netherlands, to enact new laws or legislative provisions covering genetic data and privacy. Canada has yet to join this trend. Moreover, since no genetic testing cases have been decided by high level courts in Canada, the precise degree of Charter protection of genetic privacy remains uncertain.

Equality

“Genetic discrimination” restates an old proposition: that respect for human dignity militates against discrimination based on biological status, race, religion, gender, age and disability.

Liberty

Canadian courts have recognized that the explicit protection of “liberty and security of the person” in the Canadian Charter generally protects individual rights to accept or reject governmental medical interventions. This parallels the general right to informed consent in health law.

Health

Modern human rights encompass a fundamental right to health, which could mean:

- reasonable access to genetic testing services
- participation in defining the genetic testing research agenda
- a legal requirement that testing not be made generally available until there is evidence that a test is not harmful or ineffective

Property

Some, holding the view that a person should exercise primary control over one’s genetic information, have begun to formalize “genetic property” as a human right. As well, genes for testing late onset diseases are patentable and confer exclusive property rights under federal patent law.

Public Process Norms and Values

The process side of law has played an influential role in fostering genetic testing in other countries. Inclusiveness, accountability, transparency and citizen participation are increasingly regarded as fundamental human rights norms. 

Lessons Learned

from a

British Columbia Case Study

Shahrzad Sedigh, Program Policy, Transfer Secretariat and Planning, First Nations and Inuit Health Branch, Health Canada, and Doris Cook, Policy, Planning and Priorities Directorate, Health Policy and Communications Branch, Health Canada

There is an important sense in which genetics and genomics may become “bellwethers” for issues that have been in play in other areas. For example, it is not an accident that the American federal research ethics apparatus was put under the microscope in the wake of a death in gene therapy. As the boundaries of medical research are pushed into sensitive areas of individual, family and community life, there is an obligation to reflect on the implications of this type of research for the evolution of both the research enterprise and society itself.

Often, the most critical issues are the product of a “snowball effect” in which circumstances combine over time to create risk to human health and dignity, even legal risk. Consider, for example, the interface between the social sciences and biomedicine. Many social sciences (e.g., demography, anthropology, psychology) may have an interest in the story told by the biochemical markers identified through genetics and genomics. However, the practitioners in these fields may not have the background to understand the protective protocols that apply in biomedicine. Even the protocols themselves may prove inadequate.

A recent case involving a British Columbia First Nations community illustrates some of

these “snowball effects.” The population in the community had elevated levels of rheumatic disease and experts were called in to undertake a health assessment with a major genetic element. Residents consented to the collection of tissue samples in anticipation of results that would assist with their health issue. Years passed, the original research findings were found to

be not definitive, but the tissue samples travelled to and from universities across the world without the permission of the residents of the community. This created a major issue of trust in the community, especially as some information that might have assisted residents with their health concerns was not forthcoming. Moreover, the samples, which contain important genetic information, have now entered the “global common.”

It should be noted that many of the most worrying instances of genetic research involve one or more of the following elements: vulnerable populations; sample expropriation; cultural differences that make miscommunication more likely; expertise from several disciplines; time lapses; commercialization; international trade/transactions; multi-level inquiry using global commons such as the worldwide web and banking facilities. 🌐

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Genetics & Genomics

An Unfolding Global Revolution

Phyllis Colvin continues her interview with Bulletin editor Nancy Hamilton with a focus on placing the issues described in the Bulletin against a backdrop of global developments in this area.

Q Are other countries as challenged as Canada by genetics and genomics?

Yes. It is important to recognize that we are in the midst of a global technological upheaval. The “gene” revolution, like the “chip” revolution, is part of the knowledge-based economy that is transforming economic relationships in every country. However, the gene revolution is special. Unlike other major technological advances, it will have a direct impact on us as sentient beings, not just on the world around us.

The special nature of the gene revolution has resulted in cautious management at the global and national levels. A new standard of global decision making, as represented by the Clinton-Blair agreement, requires that information from mapping the human genome remain in the public domain, despite the massive economic implications for private markets. At the national level, sensitivity to the highly personal aspects of the gene revolution was displayed in the Clinton Administration’s decisions to ban genetic testing of employees in the federal sector and to re-examine American research ethics in the wake of a death involving gene therapy. Recently, the Bush Administration has made statements designed to reinforce personal ownership and control of genetic legacy and to support, in principle, legislation on genetic privacy.

Q What are some of the long-term policy implications of this global knowledge revolution?

First and perhaps foremost are efforts to clarify how human rights protections arising from the Second World War apply to the genetic realm. Canada, along with other countries, has been involved in the

development of UNESCO’s Universal Declaration on the Human Genome and Human Rights. UNESCO continues to have an active set of international fora on genetic policy issues. Several industrialized countries have also passed legislation or are in the throes of debate about “gene laws.”

Second, increased attention has been given to so-called “founder populations.” These are populations in which, by virtue of historical isolation or lack of mobility, the frequency of genetic disease is higher than in mainstream populations. The Icelandic parliament’s debate about how to address the genetic potential offered by its population’s long-term isolation is emblematic of this policy challenge.

Canada, too, has founder populations of interest to genetic researchers — in Newfoundland and Quebec, and among Aboriginal peoples — and there is the prospect of legislation to protect founder populations in at least one Canadian province.

Third, the gene revolution will bring a large array of new services and products, many marketed over the Internet. This presents a challenge as people will look to their health system for advice about the relevance of these services and products to their short- and long-term health. It also presents a regulatory issue as governments will have to invest in the scientific capacity to ensure that these technologies are ethical, safe and effective. One concern relates to the cost of these new technologies and how they will be used in the health care system. As it is unclear whether massive investment in these technologies is warranted or whether



they can be complemented with information about how genetic disease is likely to be expressed, the prospects for predicting impacts on the health care system remain highly uncertain.

It is important to note that genetic testing has not normally been subject to federal regulation because it has generally been deemed to be a practice of medicine and, therefore, under provincial jurisdiction or a “home brew” laboratory procedure. Currently, no medical devices for conducting genetic tests have been licensed in Canada as no applications have been received. Although genetic authorities are beginning to call for increased regulation, Medical Devices Regulations do not typically cover the complexities of genetic testing, such as imprecise knowledge about its health impacts and indirect, as opposed to direct, risk of health harm — especially psychological harm.

Finally, there is an important debate in the insurance field and, indeed, in all areas of policy supported by the actuarial sciences. Some believe that the most important aspect of the gene revolution may be the potential it offers for peering into the future, a facility that is important to insurers who are interested in managing risk across populations. The policy issues characterizing the insurance/actuarial debate are spilling over into society at large. Employers, especially in the United States, have begun to use genetic information in employment-related decision making in an attempt to reduce workplace risk and long-term costs. As a result, there are now calls for broad-based legislation to control this type of use. By extension, controls may also be required in many areas of long-term life planning including, for example, education, mortgages and investment planning.

Q *How will this revolution change the way we view health and health care, and are governments prepared for these changes?*

The gene revolution is by definition multisectoral and will require a multisectoral response. It will revolutionize many parts of medicine. It will profoundly change the drug and medical device industries. It will have implications for how we understand health gain, particularly the interaction among the health determinants. It will influence the public/private mix in health care. It will reinvent large scientific domains with the creation of new disciplines such as biodiagnostics, bioinformatics and proteomics. It will

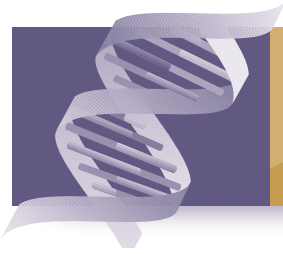
provide a new focus for technology assessment, especially as new technologies are weighed against other technologies and health enhancement strategies. Ultimately, it will change the emphasis within the clinical paradigm from addressing a disease to addressing a predisposition.

It would be fair to say that no national government is fully equipped to deal with the revolution, although the United States, Britain and Scandinavia have dedicated more policy attention to it than other jurisdictions. Undoubtedly, governments will be obliged to develop new policy processes, including the application of scientific, medical, legal, ethical and social tests in policy development. A key concern will likely be the relationship between emerging policy related to this revolution and previously established policy in other areas, such as child health, assisted human reproduction, biotechnology and overall regulatory policy.

The Canadian government’s mandate implies obligations in a number of areas: building scientific capacity in emerging disciplines; developing the necessary regulatory capacity in anticipation of the range of services and products that will be generated; protection for founder populations and populations involved in genetic research; education of populations most likely to be affected by the revolution; and hands-on involvement with participating industries to guide their development from the health, legal and ethical perspectives.

Q *Can we draw any conclusions from what has happened to date?*

One major conclusion from our global experience is that no one sector has, or could have, a monopoly on developments. Another is the recognition that the gene revolution is both global and personal. To date, global authorities, such as UNESCO, and governments in the industrialized world have been the key managers of this technology. However, this will probably change given that concerns about gene policy are a major plank of the global civil society movement. There is also a strong demand in democratic societies for decision making that is transparent, ethical, inclusive and informed. Without this, there is every prospect for powerful backlashes, especially in the area of human rights. 🌐



Who's Doing What?

Genomics and Genetics Research

Due to rapid developments in the field of genetics and genomics, the network of “players” is evolving. This article helps to clarify the landscape by highlighting Health Canada’s policy research activities in this area against the backdrop of broader coordinating mechanisms.

In the Health Portfolio

Health Canada

Health Canada is one of seven federal departments that received a significant investment in 1999 to strengthen their research capacity in genetics and genomics. Following is a snapshot of Health Canada’s research activities in this area, including genetic testing for late onset diseases.

Policy Development on Genetic Testing for Late Onset Diseases

- The Expert Working Group on Genetic Testing for Late Onset Diseases is identifying the policy, legislative, research, clinical and economic issues associated with genetic testing (see page 14).
- A national survey of Canadian laboratories conducting genetic testing has just been completed and a Genetic Testing Quality Management System is being developed (see page 12).
- Health Canada’s Working Group on Public and Professional Education on Genetic Testing for Late Onset Diseases has been mandated to assess public and professional educational needs related to genetic testing. To date, the Working Group has surveyed the educational needs of health care providers and identified available resources (e.g., interactive websites, educational modules, genetic counselling aids).

The Working Group will release its findings by March 2002, for initial review and possible public consumption. Some of the reports currently available include: *Survey on Educational Activities on Genetic Testing for Late Onset Disease: Data Analysis*, Ottawa: Health Canada, March 2000; and *Principles and Lessons Learned to Develop*

and Disseminate Genetic Testing Educational Information to the Public, Patients and Primary Care Providers for Late Onset Diseases: A Review of the Literature, Health Canada, March 2000. For copies of these reports, contact: arun_chockalingam@hc-sc.gc.ca

Surveillance Strategies Using Genetic Technologies

Health Canada has traditionally undertaken surveillance activities involving the identification of genetic markers for susceptibility to infectious diseases, as well as markers for predisposition to chronic diseases in targeted populations. Applications of new genomics-based detection technologies are now being explored.

Product Safety and Efficacy


- **Development of molecular detection technologies** — Scientists at Health Canada are developing DNA-chip technologies and related methodologies for the detection of human pathogens and food-borne pathogenic microorganisms.
- **Development of new, safe and efficient vaccines** — Health Canada is expecting that vaccines produced in edible plants will soon be submitted for approval. The department is conducting internal research in order to build scientific expertise in this area.
- **Improving food safety** — The long-term safety of genetically modified foods is a major concern to many Canadians. Health Canada is conducting systematic research in order to develop better regulatory policies for such foods.

Institute of Genetics, Canadian Institutes of Health Research (CIHR)

CIHR is a federal agency that reports to Parliament through the Minister of Health. CIHR’s Institute of Genetics is one of 13 virtual institutes that, together, cover the full spectrum of health research challenges and opportunities in Canada. The Institute’s research program is directed by its Scientific Director, Dr. Roderick McInnes in consultation with the Institute’s Advisory Board. The Institute of

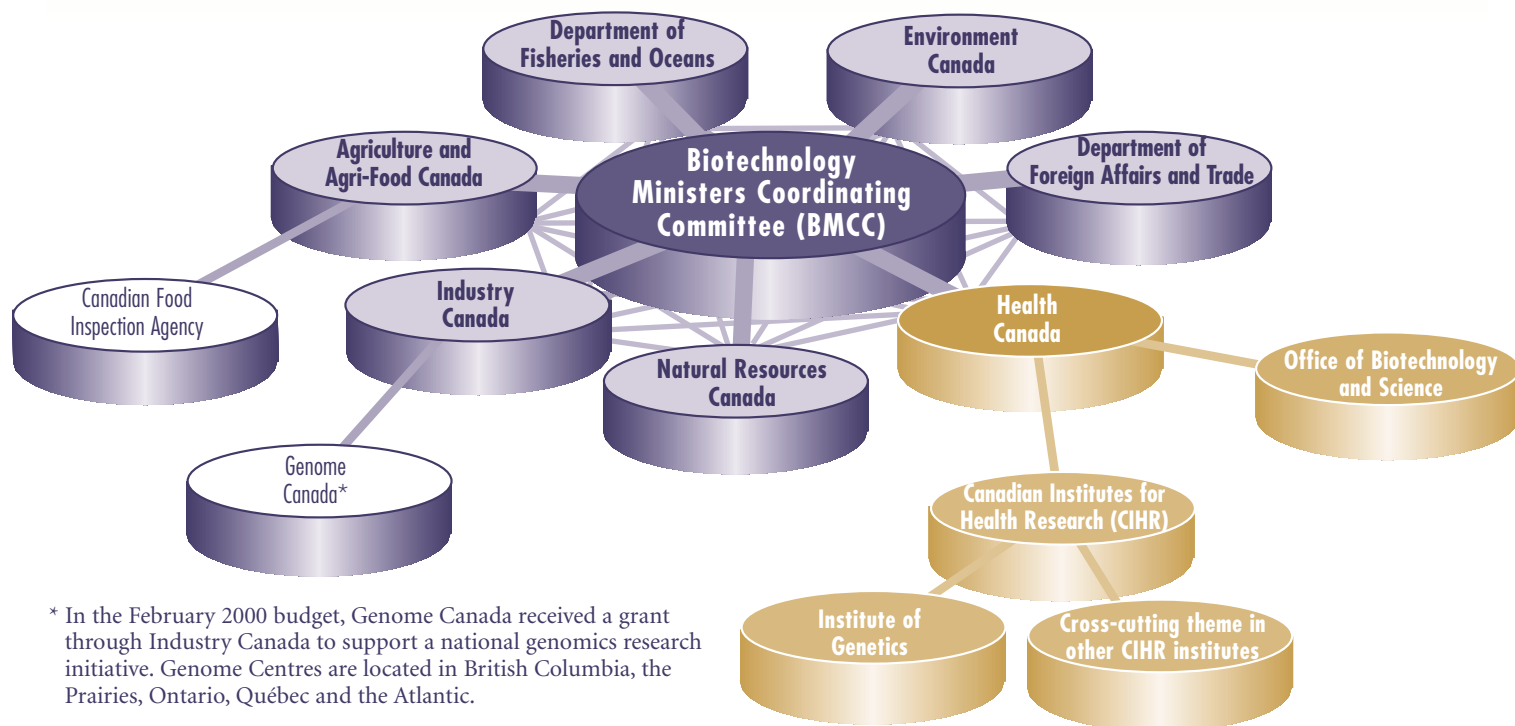
Genetics supports research on the human genome and all aspects of biochemistry and genetics related to human health and disease, including areas related to the ethical, legal and social issues of genetics research. Examples of current initiatives include the genetics of complex human diseases, population

database studies, proteomics and gene/environment interactions with human health.

Additional information on the Institute of Genetics and the Canadian Institutes of Health Research is available at <http://www.cihr.ca/index.shtml> 

The Government of Canada

The federal government has made a significant investment in developing its research capacity in the areas of genomics and genetics. The 1999 budget allocated \$55 million for improving research and development activities in this area in seven federal departments with links to the field of biotechnology. The government's activities are guided by the Biotechnology Ministers Coordinating Committee (BMCC), which was established by the Prime Minister to address biotechnology policy issues. The seven departments shown below are represented on BMCC; however, there are approximately 30 agencies and departments with an interest in biotechnology.

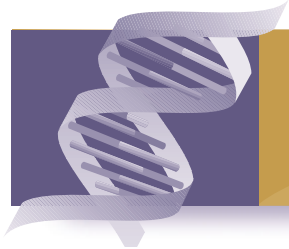


* In the February 2000 budget, Genome Canada received a grant through Industry Canada to support a national genomics research initiative. Genome Centres are located in British Columbia, the Prairies, Ontario, Québec and the Atlantic.

Canada's Biotechnology Advisory Committee

The **Canadian Biotechnology Advisory Committee (CBAC)** is an independent expert advisory committee with the mandate to advise government on policy issues related to the development and application of biotechnology in Canada. Membership on CBAC reflects a broad range of interests and expertise (e.g., health, environment, ethics, science, business, consumers).

Sithian Pandian, Office of Biotechnology and Science, Health Protection Branch, Arun Chockalingam, Centre for Chronic Disease Prevention and Control, Population and Public Health Branch, Stephanie Wilson, Health Research Secretariat, Office of the Chief Scientist, and Phyllis Colvin and Michael Silverman, Policy, Planning and Priorities Directorate, Health Policy and Communications Branch



New and Noteworthy

New and Noteworthy is a regular column of the Health Policy Research Bulletin highlighting “up and coming” policy research in the health field.

A Symposium on Genomics and Public Policy

The Government of Canada’s Policy Research Initiative is organizing a symposium that will provide researchers, industry executives, non-governmental organizations and government officials with an opportunity for high level policy discussion. Collaborators include the Canadian Biotechnology Advisory Committee, Genome Canada, the Canadian Institutes for Health Research and Health Canada. The symposium will focus on two broad, interrelated policy realms:

- **Reaping the benefits** — How can government, industry and academia work together to ensure that Canadians fully reap the health and economic benefits of genomics?
- **Stewardship issues** — What particular prevention and protection issues must be addressed if we are to benefit in a responsible and sustainable way, and how can governments act more effectively and efficiently on these issues?

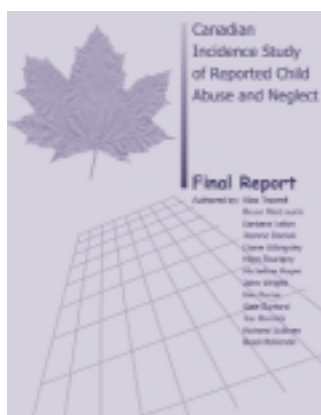
The symposium will take place between February and April of 2002, with 60-80 people participating. A number of the symposium presentations will be published in the November 2002 issue of *ISUMA: Canadian Journal of Policy Research*.

OECD Project

Because genetic testing services are often offered across borders, issues relating to genetic testing standards are international in scope. For this reason, there is an urgent need to develop internationally compatible best practice policies for analytical and clinical validation of genetic tests. The Organisation for Economic Co-operation and Development (OECD) is spearheading an international survey to measure the quality control aspects of genetic testing laboratories. Health Canada recently completed a limited survey of Canadian laboratories and will participate in the OECD survey. The Department’s Working Group on Genetic Testing for Late Onset Diseases will oversee the Canadian component of the survey. The final report is expected in 2003.

Women and Genetics

The National Network on Environments and Women’s Health, part of Health Canada’s Centres of Excellence for Women’s Health Program, has published a report entitled “The Gender of Genetic Futures: The Canadian Biotechnology Strategy, Women & Health” (NNEWH Working Paper Series, York University, Toronto, 2000). The report includes 25 papers and is based on contributions to a National Strategic Workshop organized by the Working Group on Women, Health and the New Genetics, held at York University in February 2000. The papers apply a gendered analysis to key issues in genetic testing, genetic therapies and biotechnology/genomics in general. They can be accessed at <http://www.cwhn.ca/groups/biotech/availdocs/workproc.htm> or by contacting the project coordinator at nnewh@yorku.ca



Study of Reported Child Abuse and Neglect

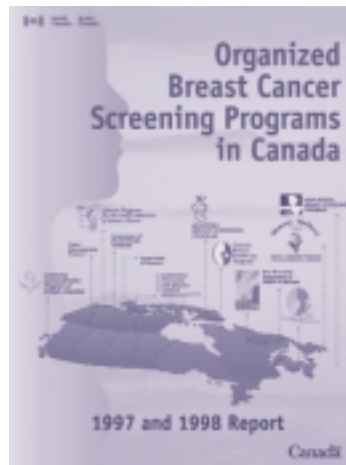
The *Canadian Incidence Study of Reported Child Abuse and Neglect* is the first nation-wide study to examine the incidence of child maltreatment in Canada. It provides comprehensive, Canada-wide statistics on children and families investigated because of suspected child abuse and neglect.

The study was a collaborative effort between Health Canada, the provincial and territorial governments, and child welfare organizations. The results have been published in three reports: *Highlights*, *Selected Results* and *Final Report*. They can be downloaded from <http://www.hc-sc.gc.ca/hpb/lcdc/brch/maltreat/index.html>

Breast Cancer Screening Programs

In December 1992, under the Canadian Breast Cancer Screening Initiative, Health Canada participated in a federal/provincial/territorial working group on breast cancer screening. The group’s mandate was to implement and evaluate breast cancer screening

programs across the country. As a result, a national database was established to monitor and evaluate breast cancer screening delivered through organized provincial programs. The second in a series of biennial reports (based on 1997 and 1998 data submitted to the database) has now been released. More information is available at <http://www.hc-sc.gc.ca/hpb/lcdc/publicat/obcsp-podcs98/index.html>



<http://www.hc-sc.gc.ca/hpb/lcdc/publicat/burden/index.html>. In addition, a semi-interactive, web-based tool provides more detailed data and additional information about data sources and methodology.

How Labour Market Experiences Contribute to Health

With funding from the Canadian Population Health Initiative, the Institute for Work and Health has addressed issues of work and health using two major national data sets — the Survey of Labour and Income Dynamics and the National Population Health Survey. The findings contribute to an understanding of workplace factors on health and how to predict future burdens of sickness and disability on work absences, productivity and pension plans. Evidence of a relationship between job status and health is likely to be a concern to employers and employees in labour market negotiations. As well, differences in the work experiences of men and women suggest that employers should consider gender in designing interventions to reduce work-related stress, illness and disability. More information can be obtained at <http://www.iwh.on.ca/Pages/Research/RAC2000/rac-area4.htm>

The Cost of Illness

A report entitled *Economic Burden of Illness in Canada, 1998* is scheduled to be released later this year. It updates and expands on information in two previous reports outlining the direct and indirect costs of illness in Canada. The data are used in health planning and priority exercises, and provide a base for ongoing work on the relative impact of health outcomes. The report, along with information on how to order copies, will be available at

Introducing Health Canada's Health Policy Working Paper Series

Health Canada's Health Policy Working Paper Series (WPS) is produced by the Applied Research and Analysis Directorate as part of a larger research dissemination program to enhance the transfer and uptake of knowledge generated within or on behalf of Health Canada. It is the first of its kind in Health Canada and will complement other Health Policy Research Communication activities, such as the *Health Policy Research Bulletin*, upcoming workshops and seminar series. The WPS will support evidence-based decision making by highlighting and promoting policy research of importance to Health Canada. All Working Papers will be available online at <http://www.hc-sc.gc.ca/iacb-dgiac/nhrdp/indexe.html>

The first five Working Papers to be published this fall will be:

1. "Pharmacare in Canada: Issues and Options" by Åke Blomqvist and Jing Xu
2. "Selected Legal Issues in Genetic Testing: Guidance from Human Rights" by Derek Jones
3. "Genetic Testing for Late Onset Diseases: Current Research Practices and Analysis of Policy Development" by Christine Jamieson
4. "Genetic Testing for Late Onset Diseases: In-depth Thematic Analysis. Policy and Jurisdictional Issues" by Christine Jamieson
5. "Immigration and Health" by Ilene Hyman

Mark Your Calendar



What	When	Theme
5th National Metropolis Conference	October 16-20, 2001 Ottawa, Ontario http://www.canada.metropolis.net/	Immigration and diversity
Canadian Public Health Association 92nd Annual Conference	October 21-24, 2001 Saskatoon, Saskatchewan http://www.cpha.ca/english/conf/92ndanl/92conf.htm	Intersectoral collaboration; healthy public policy; building capacity in vulnerable communities; improving health in Aboriginal communities
Directions IV: Advancing Health Science and the Economy	October 25-26, 2001 Toronto, Ontario http://www.parkpub.com/healthcare/information.html	Enhancing health sector innovation; exploring policies that promote health and economic competitiveness of health-related industries
Case Mix and Quality Assurance Conference: New Frontiers in Health Information	October 28-31, 2001 Niagara Falls, Ontario http://www.casemix2001.com	Case mix and quality assurance in the health care system
Caring for Health: Courageous Choices	November 4-6, 2001 Saskatoon, Saskatchewan http://www.usask.ca/caringforhealth	The influence of technology and demand; economics and access; demographics and sustainability on society and the health system
14th Health Policy Conference: Trading Away Health?	November 9, 2001 Vancouver, British Columbia http://www.chspr.ubc.ca/confere/.pdf	Globalization and health policy
2001 Innovation Conference	November 19-20, 2001 Montreal, Quebec http://www.conferenceboard.ca	Investing in innovation
Canadian Cochrane Symposium 2001	November 22-24, 2001 Edmonton, Alberta http://www.ualberta.ca/ccnc/symposium2001	"Marketing the evidence" for good health care decision making
6th International Metropolis Conference	November 26-30, 2001 Rotterdam, The Netherlands http://www.international.metropolis.net/	Migration and the cultural transformation of cities
Canadian Home Care Association 11th Annual National Home Care Conference	December 2-4, 2001 Ottawa, Ontario http://www.cdnhomecare.on.ca	A call to action — strengthening home care
2001 National Policy Research Conference: Bringing Communities Together	December 5-7, 2001 Ottawa, Ontario http://policyresearch.schoolnet.ca/nationalconference/2001/whatnew-e.html	Innovative, sustainable communities
2001 International Conference on Health Policy Research	December 7-9, 2001 Boston, U.S.A. http://www.depts.washington.edu/psasa/hpss/ichpr2001.html	Methodological issues in health services and outcomes research
10th Canadian Conference on Health Economics	May 22-25, 2002 Halifax, Nova Scotia http://www.chera.ca/	Health economics and health research