4. GENETIC HEALTH CARE SERVICES IN CANADA

The feasibility of applying genetic knowledge to medical practice, and the health care benefits that can accrue, have been demonstrated in genetic clinics across Canada for decades. The potential for benefits is greater still now that it is increasingly possible, using new DNA technologies, to analyse the genetic determinants and understand better the biological bases of disease.

Genetic technologies offer great potential health care benefits.

There are two interrelated approaches to delivery of genetic services. One approach involves integration of genetic knowledge and technologies into routine aspects of medicine. The other approach involves delivery of specialized services, usually through genetic centres.

There are two interrelated approaches to delivery of genetic services.

Some of the existing Canadian genetic services are widely respected and serve as models internationally. Examples include the maternal phenylketonuria registry and the Network of Genetic Medicine in Quebec, the British Columbia Health Surveillance Registry, and services for Huntington disease, muscular dystrophy, and cystic fibrosis patients and families.[83] Yet, despite these and other successful programs, major problems hamper the delivery of genetic health services in Canada.

Major problems hamper the delivery of genetic health services.

Genetic health services are not well integrated into Canadian health care; they remain largely isolated and treated as an esoteric medical specialty. Even the genetic centres themselves are constrained in their ability to deliver adequate services. These problems indicate that, in general, health care professionals, administrators, and decision makers do not place a high priority on the role that genes play in health, or on the contribution that genetic technologies can make in all areas of health care. Increasing cost constraints, related to rising health care costs, compound the problem.

Integration of Genetic Knowledge into Health Care

Effective genetic services will require better integration of genetic knowledge and technologies into all aspects of health care. To date, physicians and other health care practitioners have not widely incorporated genetic knowledge and tools into their practices. The potential role of genetics in health care extends far beyond the services offered at specialized genetic centres. Genetics has implications for all aspects of health care, from understanding why a specific patient has a particular disease to managing each individual's health and diseases.[84] Eventually, genetic knowledge and technologies will be among the fundamental components of health maintenance and disease control. All health care practitioners will become, to some degree, geneticists.[85]

Health care practitioners have not widely incorporated genetic knowledge and tools into their practices.

Eventually, all health care providers will become geneticists to some degree.

As the role of genetics in health and disease is integrated into medical education and related technical training, it will be possible and appropriate to decentralize testing, diagnosis, and treatment and to integrate genetics into all health care specialties, including family practice.

Genetic Centres

At present, most genetic diagnostic testing, treatment, and counselling is carried out in a few major genetic centres across Canada. Eight centres for training and service delivery are currently accredited by the Canadian College of Medical Geneticists. There are twice as many centres offering some specific genetic

More than 25 centres offer some genetic health services. Services include diagnosis, counselling, and treatment.

health services. In addition, the provinces and territories incorporate some services, including screening programs for newborns and population-specific carrier screening programs, into their general medical administration (see Appendix 3). Genetic services and patient follow-up are also sometimes handled through clinics oriented to a particular disease.

The centres offer services oriented to individuals, families, and populations.

The genetic centres offer services oriented to individuals or families, and to certain populations. Referrals to genetic centres are made for a number of reasons including occurrence in the family of disease thought to be genetic, frequent miscarriages, or abnormal fetal ultrasound results.

The referred "patients" are often families rather than individuals. Each case involves an average of 2.7 family members.[86] A Science Council survey of 10 genetic centres found that over a one-year period (1986-87) the centres dealt with almost 19 000 cases, thus treating and counselling more than 50 000 individuals.[87] Most of this activity focused on diagnosis; counselling on prognosis, risk of recurrence, and reproductive options; and treatment to alleviate symptoms and prevent complications (often long-term treatment involving support for the patient's family). Approximately 50 per cent of referrals involved prenatal diagnosis.

In 1986-87, about half of all referrals were for prenatal diagnosis...

...and only 5 per cent of lab tests were based on the new molecular DNA technologies.

In the survey period, the centres conducted more than 40 000 laboratory tests. Sixty per cent of the tests were biochemical, 35 per cent were cytogenetic, and 5 per cent were molecular. The number of molecular tests is low because the technologies are new. Only four centres analysed molecular samples, although several centres counselled patients based on results of molecular testing. Analysis of samples from population screening programs is usually done by central provincial laboratories, but genetic centres generally undertake counselling and follow-up.

The Science Council survey indicated that:

The full range of genetic technologies is not uniformly accessible to all Canadians and waiting lists for services are long.

- genetic health care technologies and services are not uniformly accessible to all Canadians;
- 80 per cent of the centres were unable to meet the current demand for laboratory and clinical services;
- waiting periods ranged from one to 10 months. Priority is given to families requiring prenatal diagnosis because the information is needed promptly;
- the administrators of Canada's health care systems have not adequately prepared for the current availability of, and the anticipated increase in, genetic technologies and the subsequent demand for services;

Genetic programs are continually struggling for funding.

- Canadian genetic programs are continually struggling for funding. Several programs are funded not through the health care system, but through research and education budgets;
- resource constraints and the priority given to prenatal diagnosis are limiting the availability of other genetic services, including treatment procedures that keep persons with genetic risk factors healthy and productive.

Over the next five years, referrals for existing services are expected to increase significantly.

Respondents at the centres estimated that over the next five years referrals for existing clinical genetic services would increase by 40 per cent, with demand for associated laboratory services increasing by 65 per cent. The demand for molecular genetic tests is anticipated to undergo the greatest growth — an increase of 160 per cent.

The centres were unable to estimate the future demand resulting from the increasing number of genetic tests that are becoming available, or the effect of the growing awareness of genetic services among the public and physicians. Among the new genetic programs that could become available over the next five years is carrier and prenatal screening for cystic fibrosis. The centres considered it unlikely that testing for multifactorial disorders would become widely available within the next five years.

Nevertheless, future demand for genetic services is likely to be substantial. In the United States it has been estimated[88] that once services become available, each year:

New tests will result in increased demand for genetic services.

- 2.7 million genetic tests will be conducted to detect young women who are carriers of cystic fibrosis, sickle cell anaemia, haemophilia, or Duchenne muscular dystrophy. It is anticipated that testing will identify 49 900 carriers. The additional testing of male partners of these carriers will identify an estimated 2800 male carriers.
- 2.4 million tests will be conducted to detect chromosome abnormalities in pregnant women/fetuses.
- when DNA-based tests become available for screening common diseases such as insulin-dependent diabetes, coronary heart disease, lung cancer, breast cancer, bipolar affective disorder, and Alzheimer disease, 16.2 million tests will be conducted to identify individuals at risk; 810 000 of these tests will be positive.

The 10 genetic centres surveyed identified several constraints that hamper their ability to deliver services effectively. Chief among these constraints is a lack of resources, trained personnel, and awareness on the part of health care providers and planners.

Lack of Funds

It is difficult to estimate the resources allocated to genetic centres because the mechanisms and sources of funding vary. A total annual operating budget of $10 million was estimated for the 10 centres surveyed by the Science Council.

Few genetic centres are funded entirely by the health care system; their funding derives from a mixture of research and education as well as health care dollars. The centres experience particular difficulty in obtaining funds to deliver verified new technologies. In addition, little funding is available for purchase of capital equipment or evaluation of new and proposed services. Capital purchases are generally funded through hospital budgets, although provinces sometimes fund the start-up costs of new or expanded programs separately.

The centres experience particular difficulty in obtaining funds to deliver verified new technologies.

Salaries of non-physician staff members at genetic centres are usually paid through operating budgets. Physicians providing services in the centres surveyed were paid from a variety of sources including provincial medical insurance programs (fee-for-service), universities, provincial genetics programs, and hospitals. Many of the geneticists surveyed felt that fee-for-service payment was unsuitable to the nature of the work and not conducive to the integration of genetic practices into other health care specialties. This form of financing does not accommodate the amount of time required to develop genetic histories, to perform and interpret tests, and to provide counselling

Fee-for-service funding hampers integration of genetics into health care.

Box 18
Available and Needed Personnel for Delivery of Genetic Health Services

The 1985 manpower survey conducted by the Canadian College of Medical Geneticists (CCMG) recognized an immediate shortfall of 34 full-time-equivalent (FTE) positions. Canadian health care personnel delivering genetic services consisted of:

- 43 FTE medical geneticists;
- 40 FTE "other professional staff."

The CCMG estimated the personnel required to provide adequate genetic services at:

- 65 FTE medical geneticists;
- 52 FTE other professionals.

The survey also predicted that an additional 43 FTE positions would be necessary by 1991. The need for trained medical geneticists and other genetics professionals will continue to grow rapidly as genetics-related technology and health care applications increase.

services for the patient and the patient's family. The average time required for a genetic consultation is three hours spread over two or more sessions.

Lack of Trained Personnel

Another limitation to service delivery is the lack of individuals trained in applied human and medical genetics. At present, the number of medical geneticists and genetics associates in Canada falls considerably below a World Health Organization recommendation[89] that one person with professional training in medical genetics be available for every 200 000 people in the population (see Box 18). The guideline itself was established before most of the new technologies, such as prenatal diagnosis, became available, and is now considered an underestimate.

Service delivery is also limited by a lack of medical geneticists and genetics associates.

There is a particular need for genetics associates. Associates are graduates of a master's-level professional program that is accredited by the Canadian College of Medical Geneticists. Working as part of a genetics team, they assist in the specialized, labour-intensive work involved in diagnosis and counselling. Although the number of available genetics associates is growing, it is not growing quickly enough because there is only one training program in Canada (at McGill University).

The delivery of genetic services and the training of personnel are complicated by the large and increasing number of identified genetic disorders and syndromes. There is a need for more computer-based information systems such as POSSUM (Pictures of Standard Syndromes and Undiagnosed Malformations),[90] both to help in the education of health care practitioners and to provide ongoing assistance in the diagnosis of genetic diseases. Such systems correlate a keyed-in description of symptoms with the disorders or mutations that could be their cause.

There is also a need for more computer-based information systems to assist in training and diagnosis.

Referral

Referral rates at genetic centres are far lower than the known incidence and risk of genetic disease would lead us to expect, and many clients are self-referred. In general, links between medical genetics and other health care disciplines at professional and administrative levels are not well established. For example, young patients with ischaemic heart disease are often not referred for genetic evaluation — a missed opportunity for preventing heart attacks in the next generation. The limited use of prenatal testing for Down syndrome also illustrates the point. The test is sensitive and can detect more than 99 per cent of cases. However, no more than 60 per cent of high-risk mothers (those 35 years of age and over) receive the test, and delivery of the service varies greatly from province to province. In a significant number of cases, the test is not offered to high-risk mothers. The reason that some doctors do not mention the procedure to their clients may be related to lack of knowledge, concern over the risks of the amniocentesis procedure, or their personal feelings on prenatal diagnosis and pregnancy termination.[91] Physicians should be aware of the role of genetics in health care, and of their responsibility to advise their patients of the existence and availability of genetic services.

Referral rates are lower than expected and many clients are self-referred.

Box 19
Health Expenditures in Canada

The value Canadians attach to their health care system is reflected in the magnitude of the resources it consumes.

- In 1987, public and private expenditures on health care in Canada totalled $47.9 billion or $1869 for every Canadian. This amount constituted about 9.0 per cent of the gross national product (GNP). Although absolute expenditures on health care have increased at a rate exceeding inflation, the proportion of GNP spent on health care has remained relatively stable since 1982.
- Canada falls just above the average among OECD countries in terms of resources spent on health care as a percentage of the gross domestic product (GDP). For example, in 1984 the United States spent approximately 10.7 per cent of its GDP on health care. The corresponding amounts for Canada and the United Kingdom were 8.4 and 5.9 per cent, respectively.
- Since 1975 approximately 75 per cent of total health expenditures in Canada every year have been publicly financed.
- Provincial governments currently spend 25-35 per cent of their budgets on health care.

Institutional health care dominates resource allocations.

- In 1987, the operating expenditures of hospitals accounted for about 40 per cent of total health expenditures and other institutions consumed another 12 per cent. Hospitals receive 90-95 per cent of their funding from government sources.
- Professional services consumed 23 per cent of national health expenditures. More than two-thirds of payments for professional services went to physicians.
- The remaining health expenditures were split between a large number of categories, including drugs and appliances (14 per cent) and public health services (4 per cent).
- Less than 1 per cent of total health resources were expended on research. This percentage has remained almost constant since 1970.

Box 20
Genetic Disease and Health Care Services: Opinions of Non-Governmental Organizations

Major findings from the Canadians for Health Research survey of 47 non-governmental organizations:

- There was wide recognition of the role of genetics in the disorder the groups represent or deal with.
- Current genetics-related programs and services were perceived as inadequate or inconsistent.
- The groups identified a need for more genetics-related health services, screening programs, counselling, and family support.
- More than 50 per cent of the organizations identified the need to address ethical issues associated with genetic research or services.
- A need for more research, including targeted research, and research funding was identified.
- There was wide agreement that better education of the public and health care providers on genetics in health and health care is needed.
- Many organizations believe they can play a greater role in advancing the "genetic point of view" in health care.

Planning

The need for effective planning for genetic services is critical as a consequence of rising demand for a growing number of services, and increasing competition for health care resources. The shortfall between supply and demand of services is increasing in all provinces.

The need for effective planning is growing.

Five provinces — British Columbia, Alberta, Ontario, Quebec, and Newfoundland — have established provincial advisory committees on genetic services. The committees vary in format and responsibilities, but essentially they review proposed technologies, the need for genetic services, and the services and budgets of provincial genetic centres. The committees assist in the efficient and rational allocation of services and funds, and they have had some success in developing genetics programs and ensuring efficient, coordinated services. There is a need for such committees in all provinces and territories.

Five provinces have established provincial advisory committees on genetic services.

The future delivery and growth of effective genetic services will depend, in part, on the development of new technologies. But the ability of the advisory committees and other interested groups to make an effective case for services and resources will also play a crucial role. Allocations will be strongly influenced by the economic climate, how the ethical issues associated with genetic technologies are handled, and the degree of awareness among the public, the health care community, non-governmental organizations, and other advocacy groups.

Future delivery and growth of services will rely on the development of new technologies and stakeholder support.

Economic Considerations

Canadians value their publicly funded health care systems but express growing concerns over the quality and accessibility of health care. At the same time, governments are concerned over costs. The rising cost of health care has resulted in a strong push for cost containment, and is making it increasingly difficult to obtain funds for new or preventive technologies. This is the health care environment into which the developing genetic technologies are being introduced (see Box 19).

Public health care systems must balance quality, accessibility, and cost.

Partly as a result of these cost restraints, the principle of accessibility to publicly funded health care is not being upheld for individuals with genetic disease. And yet some services for the prevention of genetic disease use fewer resources than the alternative treatment services.[92] For example, the provincial programs for newborn screening for hypothyroidism and the related treatment to prevent retardation are less expensive than the long-term care of individuals whose disease is diagnosed late.[93] However, at present there is no evidence that, overall, preventive genetic technologies will reduce the costs of health care. All that can be said for now is that genetic technologies have the potential to contribute to better health of Canadians (through better disease prevention and treatment) and to more cost-effective use of health care resources.

Genetic technologies have the potential to contribute to better health of Canadians and to more cost-effective use of health care resources.

The current and developing genetic technologies represent an investment in the health of Canadians that should be considered by those who make funding decisions. Failure to integrate technologies and services within publicly funded health care systems may foster the development of a two-tier system based on ability or inability to pay for privately offered services.[94]

The problems encountered in obtaining funding for genetic health services focus attention on the need for decisions on the role and funding of Canadian health care systems. Public participation in this process is essential.

Role of the Private Sector

At present, the private sector does not play an important role in the delivery of genetic services.

At present, the private sector does not play a significant role in the delivery of genetic services. Private sector labs could offer genetic analysis in the same fashion as radiology services are currently offered, with the physician acting as contact point for referrals and results.

Private sector participation is desirable if the services are publicly funded and meet appropriate standards.

Private sector participation in the delivery of genetic health services is desirable, but there are two specific concerns. If the services are not publicly funded and are available only by purchase through the private sector, the equity of Canadian health care systems would be compromised. In addition, uncontrolled private sector involvement could result in the introduction of technically unsound or inappropriate technologies and services. To ensure adequate standards, it is important that appropriate guidelines and mechanisms for provincial licensing and monitoring of facilities and services be in place. These guidelines and mechanisms should apply to both the private and the public sector.

Role of Non-Governmental Organizations

Non-governmental organizations play an important role in Canadian health care.

Non-governmental organizations (NGOs), which represent individuals and address problems related to specific disorders, have an important role in Canadian health care. These groups, such as the Huntington Society of Canada, the Canadian Cystic Fibrosis Foundation, and the Multiple Sclerosis Society of Canada, provide funding for research, are strong advocates in their dealings with health ministries and professional organizations, and offer services relevant to specific disorders. Many NGOs provide excellent social and psychological help to individuals and families with genetic diseases.

The NGOs have identified a need for more genetics-related research and health services.

Canadians for Health Research, a voluntary association, recently surveyed 47 of these organizations, representing 185 000 citizens, for their views on available services and related needs in the area of genetic disease.[95] The survey results identified the need for more genetics-related research and health services (see Box 20).

Together, the NGOs represent a large, informed constituency of researchers, individuals with genetic diseases, and health care advocates with the ability to have a significant impact on health care policy.