Survey of availability of genetic testing across Canada
Canadian College of Medical Genetics Clinical Practice Committee Document
Perspective and Recommendations

The purpose of this document is to provide information in support of equitable access to clinically relevant genetic testing to all Canadians across provinces.

Introduction: Rapid increase in molecular genetics knowledge has brought the medical community a greater understanding of the etiologies and altered physiology of a growing number of genetic conditions. Alongside this increased knowledge, advances in diagnostic technologies and availability of molecular testing also underwent a tremendous expansion.

Genetic tests are used to determine if a person has or is at risk of developing a genetic disease, or of passing on a genetic condition to an unborn child or newborn. The availability of these highly specialized investigations has changed the management of patients and families affected with genetic disorders. The aims and benefits of these tests are manifold, as described below through a few examples.

Pre-symptomatic testing: Based on family history, some Canadians are at high risk of developing cancer or a neurodegenerative disease. Examples of these conditions include inherited susceptibility to both breast and ovarian cancer (BRCA), to colon, uterine and other malignancies (hereditary non-polyposis colon cancer syndrome or HNPCC) and Huntington disease. For a person with a family history of one of these diseases, in many instances undergoing genetic testing allows that person to definitively determine whether he or she is at risk of developing the health problems that are features of the syndrome. Results of testing serve to relieve uncertainty about disease status, provide information about appropriate lifestyle choices, and influence disease management by permitting access to treatment options, services and support. In some cases the test results are used for reproductive planning and influence insurance options.

Diagnostic testing: Genetic testing can provide confirmation of a clinical diagnosis for rare disorders when signs are atypical or in a young individual in whom expression of clinical signs is uncertain, such as in Marfan syndrome. Results of testing can alleviate the burden of undergoing investigations for alternative diagnoses. In many cases, the natural history of the disease can be improved by offering clinical screening and/or more efficient clinical management.

Carrier screening: Genetic testing may provide information to persons who are not themselves at risk of developing a genetic condition, but who are at risk of passing on a particular genetic disorder to their children, either as a result of a positive family history or by ethnic background. For example, compared with people in the general population, people of Ashkenazi Jewish descent are at higher risk of being carriers of a severe neurodegenerative disorder called Tay-Sachs disease. Two individuals who are both carriers will have no manifestations of this disease, but are at a 25% risk of having an affected child. Knowledge of carrier status permits reproductive planning for couples at risk.

Prenatal Diagnosis /Assisted reproduction: For individuals with inherited genetic conditions such as Huntington disease or for carriers of recessive conditions such as cystic fibrosis, identified mutations from a genetic test may be used for prenatal diagnosis in a pregnancy or for pre-implantation genetic diagnosis (PGD) prior to pregnancy. Both of these diagnostic techniques may be used to prevent transmission of genetic disease to future offspring. The ability to test either in pregnancy or pre-implantation provides couples with reassurance and definitive information about the health outcomes of their children prenatally.

In each of these situations, genetic test results can significantly alleviate the burden of disease for individuals, families and ultimately society.
Design and result of the survey: In 2011, the Canadian College of Medical Genetics, through its Clinical Practice Committee, surveyed centers providing medical genetics services in an attempt to determine the accessibility of molecular genetic testing across Canada. The purpose of this survey was to determine the range and type of molecular genetic testing currently available to patients across Canada. The survey was not designed to determine if testing can be performed locally, but rather to determine if patients have access to clinically relevant tests.

A total of fifteen Canadian medical genetics centers responded to the survey (79% response rate). Answers were provided as yes/no to the question of availability of molecular genetic tests, with the opportunity to add comments. Conclusions drawn from the results of the survey were as follows:

1. All centers are able to offer tests for common genetic conditions such as cystic fibrosis and Fragile X syndrome. However, in certain circumstances test costs will not be covered by the provincial medical service plan. For example, in British Columbia, out-of-province pre-conception carrier testing (referred to as “family planning” by the medical provincial plan), even with relevant family history, is not covered. In this case, a woman with a family history of Lesch-Nyhan syndrome, an X-linked disorder characterized by neurologic dysfunction, cognitive and behavioral disturbances in males, would not have access to carrier testing unless a pregnancy was in progress. She would thus be denied potential access to pre-implantation diagnosis. Likewise, a couple with a family history of Smith-Lemli-Opitz, an autosomal recessive condition associated with birth defects and developmental delay, would not have access to carrier testing prior to undertaking a pregnancy. In both these scenarios, the lack of access to pre-conception molecular testing would likely lead to significant distress for at-risk families, who would face undertaking urgent genetic testing in a context of an ongoing pregnancy.

2. The majority of centers offer tests for common genetic conditions for which testing is recommended by current clinical guidelines, but a few exceptions were noted. For example, Manitoba is the only Canadian province in which patients cannot have access to testing for the genetic colon cancer syndrome HNPCC. As well, in British Colombia, out-of-province tests for inherited cancer syndromes are typically restricted to affected individuals when investigations are first initiated in a family. Thus, a person with an unequivocal family history of HNPCC-related cancers whose first-degree affected relatives were all deceased would not have access to pre-symptomatic genetic testing, although that person’s risk of having inherited a causative mutation is 50%. Clinical surveillance which includes annual colonoscopies will be required and in this case, management cannot be modified using the genetic test result.

3. Each center surveyed followed pre-established procedures to determine if testing not performed locally should be obtained. Great variations were reported among centers regarding the ease of obtaining either test approval or funding for testing. Some centers reported significant restrictions on some common non-cancer molecular tests, approval being dependent on intended use (for example prenatal diagnosis, anticipated change in clinical management, not solely for diagnostic reasons) and/or detection rate and cost.

4. Ordering of many tests is often restricted to geneticists or other medical specialists.

5. Testing for some frequent conditions such as cardiomyopathy, and Noonan or Marfan syndrome remain problematic in several provinces. Again, this means that medical resources will be devoted to prospective clinical surveillance individuals suspected of harbouring these genetic disorders, while the result of a genetic test would likely modify clinical management.
**Conclusion:** The results of this survey clearly demonstrate inequalities in access to genetic testing across Canadian provinces. The responses support a picture of a genetic testing allocation process that is dependent on regional criteria and mostly restricted to medical geneticists or other medical specialists.

Access to molecular diagnosis of genetic conditions confers multiple benefits to human health by permitting the prevention, the accurate diagnosis and/or the appropriate management of these rare disorders. It is invaluable in healthcare decision making and routine clinical practice. Access restrictions to genetic tests will likely result in poorer health outcomes for those Canadians to whom these tests are not available.

The CCMG strongly advocates equitable access to genetic testing to all Canadians, in a spirit of fairness and more equal distribution of genetic services for society as a whole. This issue must also be considered in the context of the legal and ethical duties of care of healthcare providers involved in the provision of genetic services, as well as in relation to our country’s stated commitment to universal healthcare access.