8.1.1 Summary

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The science of genetics and its clinical applications are multidimensional as a result of the mix between genetics, the environment, social, and epigenetic factors. We are beginning to understand the role of some of the factors – for example, in childhood cancers – but the future delivery of genetic services will require much more research and better integration of genetic knowledge into healthcare.

Inherited genetic conditions are critically important in the clinical care of children. The World Health Organization has stated that up to 40% of hospital care of children may be related to children with monogenic conditions (those conditions which result from a modification to a single gene).[1] Another study reports that more than 50% of paediatric hospitalizations were children with genetic conditions or children with conditions with underlying genetic components.[2] The lack of Canadian data about the disease burden of paediatric genetics poses a major challenge considering the growing demand for services, the variation in services provided, and competition for healthcare resources. To this end, it is crucial to collect and evaluate relevant data to assess the impact of genes in combination with behavior, environment, and diet on child health.

According to the 1989 Convention on the Rights of the Child, which guides our policy and healthcare approaches in Canada, the best interests of the child are paramount. To conform to the best interests of the child, certain considerations are in order when using genetic tests. Some address the relevance of treatment and prevention and others focus on the confidentiality of children's genetic test results. If a genetic test reveals the presence of a harmful mutation and there are treatments or effective preventive measures that can be initiated during childhood, it could be clearly beneficial to the actual or future health of the child. If there is no treatment or effective preventive measures that could benefit the actual or future health of the child, there should be no disclosure of the test results (either to the child or to the parents) because it would not be in the best interests of the child. Disclosure would contravene the child's rights of confidentiality and privacy and could have psychosocial consequences.

In order to give children the "best start" possible, prenatal and newborn screening programs are needed across Canada. The goal of newborn screening is to promote infant and child health by identifying babies with treatable conditions as early as possible in order to prevent death, disability, or serious health problems. The lack of national standards for newborn screening in Canada means that access to screening is not equitable for all families across the country.

Given the rapid pace of scientific advances in the field of genetics combined with the use of whole genome sequencing tests in clinical settings, there is a need to build awareness of new genomic technologies among physicians, nurses, and other healthcare providers, as each plays a unique role in the genetics healthcare team. In addition, families require access to accurate information in order to make informed decisions. They will require timely access to clinical geneticists, genetic counsellors, help lines, and support services. Such services are essential to ensure that families considering genetic testing receive up to date information about the tests, including the benefits, their accuracy, the associated risks, and the conditions that can and cannot be identified.

The evolution of the field of genetics provides us with opportunities to enable Canadian children to reach their optimum status of health and wellbeing. It also raises a number of challenges that must be addressed – in a thoughtful and evidence-based manner – to ensure the health and rights of our children are protected.

1 World Health Organization. Genes and Human Disease. Monogenic Diseases.
http://www.who.int/genomics/public/geneticdiseases/en/index2.html

2 McCandless SE, Brunger JW, Cassidy SB. The Burden of Genetic Disease on Inpatient Care in a Children's Hospital. J Hum Genet. 2004;74(1):121–7