Abstract
The presymptomatic genetic testing of minors for late onset disease has become possible through the identification of disease genes and technical development of genetic screening. However, the testing of minors poses ethical dilemmas, especially if nothing can be done to alter the natural history of the disease. This article explores the rights of the child and his or her family in the context of the decision of whether to genetically test the child. A discussion of the current ethical-legal issues surrounding the genetic testing of minors for late onset disease is explored.

Ethical Considerations
Late onset diseases (LOD) are gene-based disorders with symptoms that typically appear in adolescence or adulthood. LODs are exemplified by the progressive myoclonus epilepsies (PMEs). The PMEs typically present early in life (between 5 and 15 years of age) and are characterized by developmental regression, myoclonic seizures, and early mortality. Four of the five most common PMEs are inherited in an autosomal recessive manner, suggesting that transmission from parental carriers is possible not only to the affected individual, but also to that individual’s siblings. The PMEs are generally diagnosed clinically, but genetic testing is also available for these five most common PMEs.

When a young person is diagnosed with a PME, the genetic testing of their siblings is especially fraught with ethical dilemmas, because the onset and subsequent mortality mainly occurs before the age of consent, unlike other late onset diseases such as Huntington’s disease (HD). Patients’ parents have a vested interest in the welfare of their children as well as the welfare of the entire family unit. Some would agree that foreknowledge of the development of a non-curable disease in another child could help the family unit mentally, financially and communally prepare for the inevitable development of the disease. Indeed, many parents already exercise their right to acquire foreknowledge of genetic disease when amniocentesis is performed to screen for Down’s syndrome. A difference is that the rights of the fetus are (legally regarded as) different than the rights of a child, especially in light of the parents’ ultimate, autonomous right to terminate the pregnancy before a given gestational age. Yet, parents still exercise significant autonomy over their children from birth to adulthood, almost exclusively for their children’s sole benefit.

In defense of the rights for parents to have their children tested, some groups argue that the right to test is supported by the fact that parents are “better able to predict the psychosocial outcomes of testing than are physicians”. A survey shows that the majority of physicians tend to agree. Balanced against the family’s autonomous right (as primary caregiver to the child) to know is the child’s right to not know. Under the United Nations Charter of Human Rights, children have guaranteed universal human rights identical to adults, and additional rights specific to children. These general rights include:

Article 3:1
“In all actions concerning children, whether undertaken by public or private social welfare institutions, courts of law, administrative authorities or legislative bodies, the best interests of the child shall be a primary consideration.”

Article 12:1
“Parties shall assure to the child who is capable of forming his or her own views the right to express those views freely in all matters affecting the child, the views of the child being given due weight in accordance with the age and maturity of the child.”

As well, the Council of Europe Convention of the Protection of Human Rights and Dignity has stated that “everyone is entitled to know any information collected about his or her health. However, the wishes of individuals not to be so informed shall be observed.” Therefore, a child’s autonomous right to not know may be abrogated if the parents are permitted to exercise their autonomous rights as primary caregivers.

Children may not ultimately benefit from the presymptomatic testing of a LOD. The lessons learned in the post-test result period
after presymptomatic testing in HD have changed our approach to the dissemination of information for conditions without a cure. Specifically, the detrimental effect of a positive genetic test result on the affected individual has been studied after an increase in poor outcomes was noted. In follow-up studies of people who were tested for the HD gene, the psychological sequelae of receiving a positive result can include severe depression and suicide. In fact, one study has shown that the suicide rate of carriers of the HD gene is four times that of the national average. It is through these experiences that the World Federation of Neurology and the International Huntington’s Association forged a consensus decision in 1989 and 1990 not to test children for the HD gene. Others have followed suit, making recommendations that testing be done only when there is a clear benefit to the child. These experiences strongly suggest that knowledge of genetic information does not always benefit the individual. Further policy regarding the appropriateness of genetic testing of children is provided by the Institute of Medicine’s report on Assessing Genetic Risks, which states that “children should generally be tested only for genetic disorders for which there exists an effective curative or preventive treatment that must be instituted early in life to achieve maximum benefit. Childhood testing is not appropriate for carrier status, untreatable childhood diseases, and late onset diseases that cannot be prevented or forestalled by early treatment.” In a similar manner, policy outlined in the Genetic Privacy Act of the United States of America (Table 1) and in Canada’s Working Group on Genetic Testing emphasizes that information should not be acquired if nothing can be done about it.

### Table 1

The Genetic Privacy Act, a proposal for federal legislation to be adopted by state legislatures, is the final report of the project entitled “Guidelines for Protecting Privacy of Information Stored in Genetic Data Banks”. It was funded by the Ethical, Legal & Social Implications of the Human Genome Project through the United States Department of Energy and it recommends the following with respect to the genetic testing of late onset disease in minors.

<table>
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<th>Sec. 141.</th>
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<td><strong>AUTHORIZATION FOR COLLECTION AND ANALYSIS OF DNA FROM MINORS</strong></td>
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<td>(a) INDIVIDUALS UNDER 16. - Except as provided in sections 131(c) and 151, the individually identifiable DNA sample of a sample source who is under 16 years of age shall not be collected or analyzed to determine the existence of a gene that does not in reasonable medical judgment produce signs or symptoms of disease before the age of 16, unless:</td>
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<td>(1) there is an effective intervention that will prevent or delay the onset or ameliorate the severity of the disease; and</td>
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<td>(2) the intervention must be initiated before the age of 16 to be effective; and</td>
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<td>(3) the sample source’s representative has received the disclosures required by section 101 of this Act and has executed a written authorization which meets the requirements of section 103 of this Act and which also limits the uses of such analysis to those permitted by this section.</td>
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Several reasons are offered to defend the decision against testing children for late onset disease. First of all, “if someone learns that the child is a carrier of a gene that predisposes the child to some condition later in life, this finding may subject the child to discrimination and stigmatization by both the parents and others who may learn of this fact.” When it is discovered that a child has a predilection of developing a LOD, this child may then be treated differently than his siblings/peers. The very least of symptoms may portend a parental focus that may interfere with the social development of the child in a dignified way, a United Nations guarantee. As well, “a child’s genetic status is the child’s private genetic information and should not be determined or disclosed unless there is some compelling reason to do so.” A child has no control over the dissemination of his or her own health information and caregivers may disclose this information to whomever they desire, be it the wishes of the child or not. Thus, the ethical principle of the patient’s right to confidentiality also plays a role in the ethical dilemma of the caregiver’s autonomy to know versus the child’s autonomy to not know, be it in the present or manifested as a future decision in adulthood. At the very least, the results of genetic testing may rob a child of the opportunity to decide for himself whether or not to pursue genetic testing as a mature minor. However, this may not be relevant to the PMEs because they are likely to have manifested already by mid-adolescence. Finally, the onset of symptoms may be completely unrelated to a family disorder, but subsequent testing may provide genetic evidence of that familial disorder. Therefore, “it is essential that these clinical features are relevant to the nature of the family disorder” before testing is carried out to prevent premature disclosure of this information.

Apart from the genetic testing of children, the genetic testing of adolescents brings a different set of ethical dilemmas to the fore. In Canada, some provinces have codified the mature minor doctrine into law. This provides for an adolescent that has sufficient capacity to appreciate fully the consequences of consenting or not consenting to the various medical options so that they may, in fact, be treated the same as an adult would. Some recommend that the adolescent should be at least 14 years old before such consideration, while others have flexibly suggested that each individual case be considered separately. The UK Advisory Committee on Genetic Testing’s report on Genetic Testing for Late Onset Disorders recommends that “while ideally testing [should] be deferred until the age of majority is reached, we recommend that they are entitled to make a personal decision on this matter after a full discussion and exploration of the issues”. This is similar to Canada’s exception for mature minors. In a significant percentage of patients with a PME, the disease will clinically manifest itself after the adolescent may be considered a mature minor and due consideration must be made in these cases. Additionally, the Policy Division, Health Policy and Communications Branch, Health Canada, cautions again that when there is no clear benefit gained by the test results, the testing should be delayed until the adolescent reaches adulthood.
Legal Considerations

A search through Canada’s federal and provincial statutes, acts, regulations and court cases revealed no legal precedence of law for the genetic testing of presymptomatic disease in minors.15 The lack of cases may reflect the novelty for such litigation or it may reflect the social norm of not seeking litigation in these cases. Regardless of the lack of cases, committees are now reporting to Health Canada and these recommendations may evolve to legislation when the political need arises.

Conclusion

Intense international scrutiny of the implications of genetic testing has forced the inclusion of new ethical and social dimensions to the once separated realm of scientific inquiry. Appropriately, the Human Genome Project (HGP) has within its mandate, included a research and reporting component designed to explore the effect of the new knowledge on society and to make recommendations to governments.10 With the knowledge of the HGP results and ethical recommendations, several governments are developing policies to reflect both these recommendations and their own social context, and they are doing so in a very public manner. Medical genetics societies (such as the Canadian College of Medical Geneticists) are also offering their own recommendations to their practitioners and are paying very close attention to the public debate with the knowledge that they do not practice in a vacuum beyond the scrutiny of society. The policies under debate are many, and the stakeholders in each issue are numerous, leading to a complex consideration of all stakeholders involved.

The ethical issues that arise in pediatric genetics are especially complex due to the difference in legal and moral obligations towards minors. The fact that the rights of minors are additionally protected under many circumstances and the fact that some of these circumstances are newly evolving makes the practice of pediatric genetics both more difficult and more interesting. Using the example of the PMEs, we have seen that the identification of an autosomal recessive gene permits the disclosure of the likelihood of future ill-health in minors who are currently well. This pre-information does not necessarily permit the minor to avoid genetic disease (at least until gene therapy lives up to its much anticipated promise). Nor does it suggest a cure, since most genetic diseases currently have neither a cure nor an effective therapy. However, information regarding one’s condition may be either therapeutic or detrimental to the receiver of news, and special consideration must be made as to the impact of such knowledge on a minor in the context of his family.

References