Genetic Testing in Children: Weighing the Issues

by Diane Myles, M.S.

Testing children at risk for genetic disorders has been a recognized part of both pediatrics and medical genetics for many years. Children may undergo genetic testing for a variety of reasons. Genetic testing may have direct implications for the health of the child, either immediately, in later childhood or in adult life. Therapeutic or preventive measures are sometimes available which could influence or alter the course of the disorder if started early. In the absence of medical benefits, genetic testing may provide critical information about the presence or absence of a disabling disease, which may be useful in making reproductive decisions and future education and career choices. In some circumstances, genetic testing may not appear to offer any benefit, but is requested by the parents or the child. Many parents feel that knowledge of a future serious disorder in their child is preferable to prolonged uncertainty.

Genetic testing in children raises many ethical, legal and psychosocial issues and concerns. Testing children for cancer susceptibility raises additional concerns and, in many cases, the issues remain unresolved and testing children remains controversial. The complex issues which must be considered when children are being tested for genetic conditions include determining the medical risk-versus-benefit ratio, devising appropriate assent mechanisms for children of various ages, and the psychological and social impact of testing on children's development. Issues of confidentiality and discrimination must also be considered. Although parents may believe that they are acting in the best interests of their child, genetic testing may have negative implications and potentially damaging effects on their own future and the child's future. Sometimes an immediate benefit, such as a reduction in uncertainty and/or anxiety, causes parents and professionals to overlook the potential harms of genetic testing. Stigmatization, including lowered expectations within the family and feelings of "survivor guilt," are longer-term psychosocial outcomes which may have implications for the functioning of the family unit. Testing may also affect the child's self concept and bonds between the child and other family members.

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It has been recommended by geneticists and ethicists in North America and the United Kingdom that genetic testing only be performed in childhood if there is clear benefit to the minor. General guidelines published by the American Society of Human Genetics (ASHG) and the American College of Medical Genetics (ACMG) suggest that timely medical benefit should be the primary justification for genetic testing in children. In recognition of the fact that many of the benefits and harms of testing are psychosocial as opposed to physical, substantial psychosocial benefit to the competent adolescent is also considered appropriate justification for genetic testing. The ASHG and ACMG feel that genetic testing should generally be deferred in situations where the medical or psychosocial benefits will not become relevant until adulthood. In cases where the balance of benefits and harms is uncertain, the decision regarding testing should be based upon the unique circumstances of each family. In such situations, it is important for the provider to facilitate a thorough discussion of the potential benefits and harms to the child and assess the family's understanding of these issues. Ultimately, however, the provider should respect the decision of competent adolescents and their families.

A major question concerning presymptomatic genetic testing is: at what age should the test be justifiable? In some conditions, such as neurofibromatosis type I (NF1), where phenotypic abnormalities sufficient to allow a clinical diagnosis are almost always present by 5 years of age, the need for predictive genetic testing is minimal. In late-onset disorders such as Huntington's disease, Alzheimer's disease and hereditary breast and ovarian cancer, where symptoms are rarely seen in childhood, predisposition testing is generally not recommended for children. In disorders that are much more variable, where pressure to test children may be greater, the decision becomes more complicated.

The potential loss of autonomy for the child is a focus of much concern in relation to predisposition testing of children. When children are tested in childhood, at the request of their parents, they lose the right to make an informed decision regarding testing when they reach the age of majority. In addition, the child loses the right not to be tested and his right to privacy. Out of respect for a child's autonomy, it has been suggested that in the absence of clear medical benefit, parents should "restrain their desire to know" and not pursue genetic testing for their children.

The term "assent" is used to distinguish a minor's agreement to parental decisions about treatment, research or testing, from an adult's legal "consent," and is based on the assumption that a child does not have sufficient autonomy

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to request or consent to a procedure independently, but does have a great enough understanding of the procedure to agree with his parents' decision. In regards to research, it has been recommended that children seven years of age or older be required to provide assent. With regards to predisposition testing, it has been suggested that it is unethical, even in the event where it is legal, for a parent to consent to predisposition testing for an unwilling child, when the child is of sufficient age to understand the issues involved. In a study designed to examine the attitudes of mothers of pediatric oncology patients towards cancer predisposition testing in children, 23% of mothers felt that children's views should be routinely sought and considered in the decision about genetic testing and 55% said that consideration of the child's wishes depended entirely upon the age of the child. Twenty-two percent of mothers stated that they would not involve their child in discussions or decisions regarding testing. The mothers voiced a wide range of opinions about the age at which a child should be involved in the decision-making process. At this point in time, issues concerning autonomy and providing assent for predisposition testing in childhood remain unresolved. To determine the age at which a child can sufficiently understand the relevant issues to participate in the decision-making process and give informed assent will involve complex analysis. Careful examination of individual risks and benefits will be necessary to protect the rights of minors.

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There are clearly some circumstances where testing children for an inherited cancer predisposition is beneficial. Familial adenomatous polyposis, von Hippel-Lindau disease and neurofibromatosis type 2 are all classical tumor predisposing conditions with variable phenotype and onset. Predictive testing for mutations in the genes which predispose to these disorders is important in the identification of individuals most likely to benefit from screening aimed at the early detection and prevention of malignant lesions. Additionally, for individuals at risk, a negative test result warrants withdrawal from sometimes unpleasant and tedious screening.

In multiple endocrine neoplasia type 2 (MEN-2), testing for the inherited mutation in RET in early childhood will separate those individuals who require prophylactic thyroidectomy to prevent "C" cell tumors from those who require no further surveillance. These examples illustrate situations where testing for predisposition is clearly advantageous for the health of the child. Effective treatments and methods of early detection and prevention are in place so that learning in childhood of one's increased chances for cancer has the potential to decrease morbidity and mortality.

In Li-Fraumeni syndrome, pediatric cancers are a frequent occurrence; however, the decision to test at-risk children for the presence of constitutional TP53 mutations is not an easy one. Early on, in 1992, when mutation analysis for TP53 mutations first became available, testing children within LFS families was recommended. Since that time, many testing programs have rethought this decision. In the absence of an accepted screening program and/or effective medical intervention, benefit to the child is less easily indicated and in general, testing is not advocated in childhood.

In light of the challenging ethical issues which surround predisposition testing in childhood, the decision to test children is always very complex; however, when medical interventions exist, and the benefits of testing appear to outweigh the potential risks, parents and professionals can justify testing at a young age.

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