

Genetic Testing of Children for Adult-Onset Diseases:

Is Testing in the Child's Best Interests?

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Abstract

Questions related to testing children for adult-onset genetic diseases include many variables that generate different answers among different families. These issues include the biological nature of specific genes, concern about what benefits or harms may accrue from testing children, possible psychosocial sequelae, and ethical and legal concerns about personal autonomy. The shift in the physician-patient relationship from professional beneficence to patient autonomy has established the patient, or parents, as the primary decision-makers in questions about treatment or testing options. The role of parents as decision-makers for their own minor children has been reinforced by four seminal holdings of the United States Supreme Court. Assertions about protecting the future autonomy of children are invalid because minor children are not autonomous. Their parents, on the other hand, have a right—and perhaps even a duty—to exercise their own vested autonomy in making decisions that they believe are in the best interests of their own families. Geneticists are urged to provide clear and complete counseling to parents who seek testing for their children, and then defer to the parents as the primary decision-makers for their own minor children.

Key Words: Genetic testing, children, adult-onset disease.

Introduction

OVER THE PAST DECADE the dilemmas of genetic testing of children have generated much discussion and debate. The controversy first erupted in 1990 when two investigators in Canada asserted that no children should ever be tested for the gene that causes Huntington disease (HD) (1). The rationale offered by these investigators was based on the natural history of the disease, including the fact that HD is an adult-onset, neurodegenerative disease for which there is no treatment or cure. The autosomal dominant gene is fully penetrant, or expressed, and the disease is inevitably fatal. The authors asserted an obligation of professionals to

protect the future autonomy of the children. They further asserted that the parents of children who might carry the gene for HD should be treated as third parties with respect to their own children and that decisions about testing should rest with physicians. This last assertion placed the physician squarely between parents and their children.

The responses to the Canadian authors were at first heated and later more measured. The American Society of Human Genetics and the American College of Medical Genetics together designated a task force to examine the issues. The resulting "Points to Consider" paper stopped short of suggesting a complete moratorium on testing children for adult-onset genetic diseases. The task force carefully considered both the benefits and the harms that could result from testing children, including both medical and psychosocial consequences for children as well as their parents. The task force urged thorough genetic counseling for the parents, with the health care provider acting as the advocate for the children's interests. The tone of the joint paper was decidedly cautionary, with clear admonitions to both parents and professionals to consider the options and possible consequences of testing and of using the information generated by testing (2).

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This paper reviews the salient issues in the debate about testing children for genes that cause diseases of adulthood. These issues include the many variables that come into play in making decisions about testing children, recent shifts in the balance of the physician-patient relationship, and the roles of parents, children, and professionals in the practice of genetic testing and genetic counseling.

Variables in Genetic Testing

Decisions about testing children for genes that cause adult-onset diseases are based on a complicated and evolving set of variables. The first and fundamental set of variables relates to the biological nature of the disease of interest. Is the disease caused by a single gene that is readily identifiable, or is the disease the result of a complex interaction of hereditary and environmental factors that are difficult to define and document exactly? If the disease is caused by a single gene, is it caused by a single dominant allele or by a pair of recessive alleles? Has previous research yielded reliable information about the penetrance and variable expressivity of the gene, or genes, in question, and the risks associated with reduced penetrance or a wide range of expression? Is the disease amenable to environmental or medical manipulation? What is the natural course, or severity, of the disease? Answers to these questions will provide basic information about the disease itself and possible pathways for managing the health care of persons who develop the disease (3).

A second set of variables involves the possible costs and benefits that might accrue from testing children for adult-onset diseases. Parents who face the appearance of genetic disease in their offspring may seek to arrange their financial affairs to provide for the inevitable expenses related to medical care and treatment. Indeed, parents may wish to provide trusts for children who will later be affected, so that their unaffected offspring will not be burdened with the costs of caring for affected siblings. Further, parents may wish to relocate their families to centers of specialized research and care for persons who will later develop a difficult condition or disease. While most geneticists agree that providing a medical benefit for children who are tested is the overriding reason for such testing, we must acknowledge that financial and other factors can be of great importance in dealing with the burden of genetic disease.

Geneticists have long recognized the significance of the psychosocial consequences of generating genetic information about children and adolescents. How this information is generated and

how it is imparted after testing can have either positive or negative consequences. If the information is carelessly conveyed to children, they may experience depression or loss of self-esteem, and there may be psychiatric sequelae. Children may develop "survivor guilt" based on the knowledge that they may outlive their affected siblings or they may experience a sense of identifying with the family member who will be among the affected. These factors must be taken into consideration when counseling parents both before and after testing, and the discussion should include the possibility of not revealing test results to children at all (4).

A final variable in deciding whether to test children for late-onset diseases is the question of personal autonomy. Some children mature more slowly than others. Children reach autonomy, or the right to make their own decisions, only when they reach the legal age of majority. The child's parents have the legal right to make major decisions for the child until the legal age of majority, with the exceptions of the legal emancipation of a minor or in cases of illegal abuse or neglect. While some professionals claim the right to protect the child's future autonomous interests, they stand at odds with the parents, who should and must exercise their own vested autonomy in making decisions on behalf of their own families.

The Physician-Patient Relationship

The relationship between physicians and patients has evolved significantly over the past several decades. The fiduciary nature of the relationship has traditionally been based on an inherent imbalance between the physician's special training and expertise in medicine and the patient's lack of knowledge, requiring the patient to place considerable faith in the physician's judgment and recommendations. In recent decades, however, as patients have become more curious and informed about their health care, the basis of decisional authority in the relationship has shifted from the physician to the patient. This shift has redefined the physician's fiduciary duty, from a duty to make the best decision for the patient, to a duty to inform the patient about various options so that the patient can make a fully informed decision for himself, herself, or for offspring. Indeed, some professionals in medical genetics have further argued that "[i]n genetics, the patient is really the family rather than the individual, which means that duties to relatives may take precedence over individual rights to privacy" (5).

Roles of Parents, Children and Geneticists in Genetic Testing

Over much of human evolution, parental authority has helped maintain the family as the basic social unit. While the authority of the father over the family was traditionally predominant, women and children have gradually attained the rights of personhood in the eyes of society and the law, and they are no longer regarded as chattel, or property, subject to sale or even death, at the judgment of the husband or father (6). In the United States, in the 20th century, four seminal cases in constitutional law have reinforced the right of parents—fathers and mothers—to make decisions for their own families. The Supreme Court affirmed that the meaning of constitutional “liberty” interests can be extended to include “the right of the individual to contract, to engage in any of the common occupations of life, to acquire useful knowledge, to marry, establish a home and bring up children” (7). A second ruling of the Court found a “liberty of parents and guardians to direct the upbringing and education of children under their control.... The child is not the mere creature of the State; those who nurture him and direct his destiny have the right, coupled with the high duty, to recognize and prepare him for additional obligations” (8). A third case affirmed the right of Amish parents to school their own children after eighth grade, holding that denying this right might create “such an intrusion by the State into family decisions in areas of religious training” that might raise “grave questions of religious freedom” (9). Finally, in the most significant case that arose from the Baby Doe case of the 1980s, the Court affirmed that “[s]tate law vests decisional responsibility in the parents, in the first instance” (10). With these rulings as a foundation, both society and the law give parents wide latitude in their roles as caretakers and decision-makers for their own children, and both society and the law operate from the presumption that parents act in the best interests of their children. These arguments are readily extended to support the right of parents to seek genetic testing for their own children, including testing for adult-onset diseases.

In addition to affirming the role and rights of parents, society has also made significant progress in establishing and protecting the rights of children. The law entitles children to adequate care and the right to live free from abuse or neglect. These rights are supported by both civil and criminal sanctions for violations (11). The value of children in our society is further acknowledged by immunization and other health care programs, by

public education and recreation programs, and by “parenting” programs to help parents through the rigors of raising and caring for children. Further appreciation of the rights of children is now evidenced by the tendency to seek the “assent” of more mature children to receive medical treatment or to participate in research protocols.

An important area of concern in the relative roles of parents and children derives from the principle of personal autonomy, or the right to make one’s own decisions. Children are not autonomous persons in the law until they reach the age of majority or are legally emancipated. Parents, on the other hand, are by definition autonomous persons; this status is attained either by reaching the age of majority or by legal emancipation following the birth of a child. A significant consequence of parental autonomy is parents’ right to make decisions that they believe to be in the best interests of their own children and families. These decisions may include having a child tested for late-onset genetic diseases, thereby allowing both parents and children to explore appropriate options for financial planning and medical management.

Just as the roles of parents and children continue to evolve in applications of our new genetic technologies, so does the role of geneticists change in questions of testing children for late-onset diseases. One of the classic goals of genetic counseling is to help patients “choose the course of action which seems appropriate to them in view of their risk and their family goals and [to] act in accordance with that decision” (12). Viewing the family as the patient, the geneticist helps the family make the best possible adjustment to its genetic situation in view of its values and circumstances. These adjustments may include, for example, decisions to avoid further pregnancies, or decisions to pursue alternative options for having children.

Genetic counseling is often complex and time-consuming, and ideally it is individually tailored to each patient or family. Indeed, counseling is a *process* that may involve several sessions, so that both the counselor and the patients can be assured of a common understanding of all relevant issues. Providing several counseling sessions allows for a flexible protocol and reiteration of points of information that may be reviewed and clarified, as needed, from one session to the next. As well as being an extended process, the *content* of genetic counseling should include information about both the positive and negative implications and consequences of testing children, with thorough discussion about the role of parents in disseminating—or not disseminating—genetic information to children and other family members (13).

Conclusions

Is testing children for adult-onset genetic diseases in the children's best interests? In view of the issues discussed above, the answers to this question become complex and will vary from one family to the next. In general, however, genetics professionals are wise to avoid generating across-the-board rules that may not be in the best interests of the child in all situations. Geneticists should be continually aware that each family represents a unique set of circumstances. The obligations of the geneticist are to present genetic information and its implications in a thorough manner and in understandable language, and to explore with parents the possible options for the family. After clear, careful, and thorough counseling, the genetics professional should defer to the parents as the primary decision-makers for their own families. The final caveat helps the genetics professionals who may occasionally find themselves in situations that are ethically questionable: while professionals are never required to cooperate with requests or activities that are morally repugnant, they may well have an obligation to refer the parents or family to another professional who may be more sympathetic to testing children for adult-onset diseases.

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