ASHG/ACMG REPORT
Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents

The American Society of Human Genetics Board of Directors and The American College of Medical Genetics Board of Directors

Rapid developments in genetic knowledge and technologies increase the ability to test asymptomatic children for late-onset diseases, disease susceptibilities, and carrier status. These developments raise ethical and legal issues that focus on the interests of children and their parents. Although parents are presumed to promote the well-being of their children, a request for a genetic test may have negative implications for children, and the health-care provider must be prepared to acknowledge and discuss such issues with families.

This report is grounded in several social concepts: First, the primary goal of genetic testing should be to promote the well-being of the child. Second, the recognition that children are part of a network of family relationships supports an approach to potential conflicts that is not adversarial but, rather, emphasizes a deliberative process that seeks to promote the child’s well-being within this context. Third, as children grow through successive stages of cognitive and moral development, parents and professionals should be attentive to the child’s increasing interest and ability to participate in decisions about his or her own welfare.

Counseling and communication with the child and family about genetic testing should include the following components: (1) assessment of the significance of the potential benefits and harms of the test, (2) determination of the decision-making capacity of the child, and (3) advocacy on behalf of the interests of the child. The following points should be considered:

I. Points to Consider

A. The Impact of Potential Benefits and Harms on Decisions about Testing

1. Timely medical benefit to the child should be the primary justification for genetic testing in children and adolescents. Under this condition, genetic testing is similar to other medical diagnostic evaluations. Medical benefits include preventive measures and therapies, as well as diagnostic information about symptomatic children. If the medical benefits are uncertain or will be deferred to a later time, this justification for testing is less compelling.

2. Substantial psychosocial benefits to the competent adolescent also may be a justification for genetic testing. The benefits and harms of many genetic tests are psychosocial rather than physical. Relevant issues include anxiety, self-image, uncertainty, and the impact on decisions relating to reproduction, education, career, insurance, and lifestyle.

3. If the medical or psychosocial benefits of a genetic test will not accrue until adulthood, as in the case of carrier status or adult-onset diseases, genetic testing generally should be deferred. Exceptions to this principle might occur when the adolescent meets conditions of competence, voluntariness, and adequate understanding of information. Further consultation with other genetics services providers, pediatricians, psychologists, and ethics committees may be appropriate to evaluate these conditions.

4. If the balance of benefits and harms is uncertain, the provider should respect the decision of competent adolescents and their families. These decisions should be based on the unique circumstances of each family. The provider should enter into a thorough discussion about the potential benefits and harms and should assess the family’s understanding of these issues.

5. Testing should be discouraged when the provider determines that potential harms of genetic testing in chil-
dren and adolescents outweigh the potential benefits. A health-care provider has no obligation to provide a medical service for a child or adolescent that is not in the best interest of the child or adolescent.

B. The Family's Involvement in Decision Making

1. Education and counseling for parents and the child, commensurate on maturity, should precede genetic testing. Follow-up genetic counseling and psychological counseling also should be readily available. Providers of genetic testing should be prepared to educate, counsel, and refer, as appropriate.

2. The provider should obtain the permission of the parents and, as appropriate, the assent of the child or consent of the adolescent. Decisions about competence should not depend arbitrarily on the child's age but should be based on an evaluation of the child's cognitive and moral development. The provider should also attempt to establish that the child's decision is voluntary.

3. The provider is obligated to advocate on behalf of the child when he or she considers a genetic test to be—or not to be—in the best interest of the child. Continued discussion about the potential benefits and harms—and about the interests of the child—may be helpful in reaching a consensus.

4. A request by a competent adolescent for the results of a genetic test should be given priority over parents' requests to conceal information. When possible, these issues should be explored prior to testing. When a younger child is tested and the parents request that the provider not reveal results, the provider should engage the parents in an ongoing discussion about the benefits and harms of the nondisclosure, the child's interest in the information, and when and in what manner the results should be disclosed.

C. Considerations for Future Research

As genetic testing for children and adolescents becomes increasingly feasible, research should focus on the effectiveness of proposed preventive and therapeutic interventions and on the psychosocial impact of tests. Such data are necessary to define the empirical benefits and harms of testing before judgments about the advisability of testing are formulated.

II. Discussion

Benefits and Harms of Genetic Testing in Children

Parents sometimes request that their children be tested for adult-onset problems, so that they can address psychosocial issues. Such nonmedical uses by parents are one of the most controversial issues in testing children (Working Party of the Clinical Genetics Society 1994). While some providers argue that parents should be able to obtain such information (Pelias 1991), other providers suggest that access to such information should be restricted or prohibited if the children will realize little or no immediate medical benefit (Harper and Clarke 1990). Some geneticists already limit testing for adult-onset diseases to individuals who are >18 years of age, e.g., in some protocols for Huntington disease (Bloch and Hayden 1990) and breast cancer (Biesecker et al. 1993). One justification has been that, since such testing requires informed consent, and since children are not competent to give consent, therefore children should not be tested. However, this argument is so broad that it would preclude all pediatric care.

As with any other medical intervention, when children do not have the capacity to provide voluntary, informed consent, the decisive consideration in genetic testing in children should be the welfare of the child. Decisions about genetic testing in children should be based on an assessment of the possible benefits and harms that may be associated with the tests (see table 1). The putative benefits and harms include medical, psychosocial, and reproductive issues that have implications for the child, the immediate family, and more distant relatives.

Medical issues.—Medical issues include the possibilities of treatment and prevention, decisions about surveillance, and the resolution of questions about prognosis and diagnosis.

1. Treatment and prevention. Tests that offer children the potential for therapeutic benefit are most likely to be supported by the public and by medical professionals. For example, testing for familial hypertrophic cardiomyopathy, a disease associated with increased risk for sudden death, allows drug therapy to prevent arrhythmias (Maron et al. 1987). Individuals identified as having genetic diseases or disease susceptibility may also benefit from preventive advice about lifestyle changes. For example, children with familial hyperlipidemia may benefit from dietary restrictions (Cortner et al. 1993).

Although some medical benefits from diagnosis in childhood are established, others remain unconfirmed—and may even be associated with the possibility of harm. One possible harm to a child determined to have a deleterious gene is increased medical tests and treatment regimens that may not have proved benefits. For example, presymptomatic diagnosis of cystic fibrosis has not yet demonstrated any medical benefit and may be associated with increased costs, unnecessary treatments, and familial distress (Farrell and Mischler 1992). Thus, the potential for benefit of unestablished treatment and/or prevention regimens is a questionable justification for testing. Empirical verification of the benefits and harms of prevention and treatment should precede recommendations for routine testing (Wilfond and Nolan 1993; Marteau 1994).

2. Surveillance. Genetic testing can identify patients