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Ethical and Policy Issues in Genetic Testing and Screening of Children
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POLICY STATEMENT

Ethical and Policy Issues in Genetic Testing and Screening of Children

abstract

FREE

The genetic testing and genetic screening of children are commonplace. Decisions about whether to offer genetic testing and screening should be driven by the best interest of the child. The growing literature on the psychosocial and clinical effects of such testing and screening can help inform best practices. This policy statement represents recommendations developed collaboratively by the American Academy of Pediatrics and the American College of Medical Genetics and Genomics with respect to many of the scenarios in which genetic testing and screening can occur. *Pediatrics* 2013;131:620–622

BACKGROUND

In 1953, Watson and Crick described the DNA double helix. Fifty years later, the full sequence of the human genome was published. Our knowledge of genetics grows rapidly, as does consumer interest in undergoing genetic testing. Statements about genetic testing of children in the United States written in the past 2 decades need to be updated to consider the ethical issues arising with new technologies and expanded uses of genetic testing and screening.^{1,2} The growing literature on the psychosocial and clinical effects of such testing and screening can help inform us about best practices.

Genetic testing and screening of minors are commonplace. Every year, ~4 million infants in the United States undergo newborn screening for metabolic, hematologic, and endocrine abnormalities for which early treatment may prevent or reduce morbidity or mortality.

Outside of newborn screening, genetic testing of children is less commonly performed. Diagnostic genetic testing may be performed on a child with signs or symptoms of a potential genetic condition or for treatment decisions made on the basis of results of pharmacogenetic assays. Genetic testing may also be performed on an asymptomatic child with a positive family history for a specific genetic condition, particularly if early treatment may affect morbidity or mortality. The American Academy of Pediatrics (AAP) and the American College of Medical Genetics and Genomics (ACMG) provide the following recommendations regarding genetic testing and screening of minors. An accompanying technical report provides ethical explanations and empirical data in support of these recommendations (<http://www.nature.com/gim/journal/vaop/ncurrent/full/gim2012176a.html>).³

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GENOMICS SOCIAL, ETHICAL, AND LEGAL ISSUES COMMITTEE

KEY WORDS

genetic testing, genetic screening, newborn screening, predictive testing, disclosure, carrier identification

ABBREVIATIONS

AAP—American Academy of Pediatrics

ACMG—American College of Medical Genetics and Genomics

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GENERAL RECOMMENDATIONS

1. Decisions about whether to offer genetic testing and screening should be driven by the best interest of the child.
2. Genetic testing is best offered in the context of genetic counseling. Genetic counseling can be performed by clinical geneticists, genetic counselors, or any other health care provider with appropriate training and expertise. The AAP and ACMG support the expansion of educational opportunities in human genomics and genetics for medical students, residents, and practicing pediatric primary care providers.

DIAGNOSTIC TESTING

3. In a child with symptoms of a genetic condition, the rationale for genetic testing is similar to that of other medical diagnostic evaluations. Parents or guardians should be informed about the risks and benefits of testing, and their permission should be obtained. Ideally and when appropriate, the assent of the child should be obtained.⁴
4. When performed for therapeutic purposes, pharmacogenetic testing of children is acceptable, with permission of parents or guardians and, when appropriate, the child's assent. If a pharmacogenetic test result carries implications beyond drug targeting or dose-responsiveness, the broader implications should be discussed before testing.

NEWBORN SCREENING

5. The AAP and ACMG support the mandatory offering of newborn screening for all children. After education and counseling about the substantial benefits of newborn screening, its remote risks, and the next steps in the event of a positive screening result, parents should

have the option of refusing the procedure, and an informed refusal should be respected.

CARRIER TESTING

6. The AAP and ACMG do not support routine carrier testing in minors when such testing does not provide health benefits in childhood. The AAP and ACMG advise against school-based testing or screening programs, because the school environment is unlikely to be conducive to voluntary participation, thoughtful consent, privacy, confidentiality, or appropriate counseling about test results.
7. For pregnant adolescents or for adolescents considering reproduction, genetic testing and screening should be offered as clinically indicated, and the risks and benefits should be explained clearly.

PREDICTIVE GENETIC TESTING

8. Parents or guardians may authorize predictive genetic testing for asymptomatic children at risk of childhood-onset conditions. Ideally, the assent of the child should be obtained.
9. Predictive genetic testing for adult-onset conditions generally should be deferred unless an intervention initiated in childhood may reduce morbidity or mortality. An exception might be made for families for whom diagnostic uncertainty poses a significant psychosocial burden, particularly when an adolescent and his or her parents concur in their interest in predictive testing.
10. For ethical and legal reasons, health care providers should be cautious about providing predictive genetic testing to minors without the involvement of their parents or guardians, even if a minor is mature. Results of such tests may have significant medical, psychological, and social

implications, not only for the minor but also for other family members.

HISTOCOMPATIBILITY TESTING

11. Tissue compatibility testing of minors of all ages is permissible to benefit immediate family members but should be conducted only after thorough exploration of the psychosocial, emotional, and physical implications of the minor serving as a potential stem cell donor. A donor advocate or similar mechanism should be in place from the outset to avert coercion and safeguard the interests of the child.⁵

ADOPTION

12. The rationale for genetic testing of children in biological families should apply for adopted children and children awaiting placement for adoption. If a child has a known genetic risk, prospective adoptive parents must be made aware of this possibility. In rare cases, it may be in a child's best interest to undergo predictive genetic testing for a known risk before adoption to ensure the child's placement with a family capable of and willing to accept the child's potential medical and developmental challenges. In the absence of such indications, genetic testing should not be performed as a condition of adoption.

DISCLOSURE

13. At the time of genetic testing, parents or guardians should be encouraged to inform their child of the test results at an appropriate age. Parents or guardians should be advised that, under most circumstances, a request by a mature adolescent for test results should be honored.
14. Results from genetic testing of a child may have implications for the parents and other family

members. Health care providers have an obligation to inform parents and the child, when appropriate, about these potential implications. Health care providers should encourage patients and families to share this information and offer to help explain the results to the extended family or refer them for genetic counseling.

15. Misattributed paternity, use of donor gametes, adoption, or other questions about family relationships may be uncovered “incidentally” whenever genetic testing is performed, particularly when testing multiple family members. This risk should be discussed, and a plan about disclosure or nondisclosure should be in place before testing.

DIRECT-TO-CONSUMER TESTING

16. The AAP and ACMG strongly discourage the use of direct-to-consumer and home kit genetic testing of children because of the lack of oversight on test content, accuracy, and interpretation.

LEAD AUTHORS

Lainie F. Ross MD, PhD (AAP Committee on Bioethics)
Howard M. Saal, MD (AAP Committee on Genetics)
Rebecca R. Anderson, JD, MS (ACMG Social, Ethical, and Legal Issues Committee)
Karen L. David, MD, MS (ACMG Social, Ethical, and Legal Issues Committee)

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Beth Anne Tarini, MD, MS

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Alexander Asamoah, MD, PhD
Gary S. Gottesman, MD
Lainie F. Ross, MD, PhD
* Dr Masamichi Ito works for Athena Diagnostics, a company that performs genetic testing in minors.

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