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ASHG issues position statement on genetic testing in children and adolescents

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The American Society of Human Genetics (ASHG) Workgroup on Pediatric Genetic and Genomic Testing has issued a position statement on Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents. Published today in *The American Journal of Human Genetics*, the statement aims to guide approaches to genetic testing for children in the research and clinical contexts. It also serves as an update to the Society's 1995 statement of the same title, which was issued jointly with the American College of Medical Genetics.

"Twenty years ago, genetic tests were first being introduced into clinical medicine, and they focused on single-gene disorders in the context of family history and population screening," said Jeffrey R. Botkin, MD, MPH, first author of the report and chair of the working group. "At that time, we had limited data on how genetic testing affected children and their families, and generally suggested that unless obtaining this data could provide timely medical benefits to the child, testing should be deferred to adulthood," he added.

Since then, the scope and accuracy of genetic testing have improved, and health professionals have gained experience explaining, recommending, and administering such tests. To reflect these changes, the new statement addresses a variety of issues related to genetic testing, including:

- Scientific advances such as whole-genome sequencing (WGS), chromosomal microarray analysis (CMA), and

pharmacogenomics
- Predictive testing in children for adult-onset conditions
- Ethical consideration of how to handle so-called "secondary findings" - findings unrelated to the condition that prompted the genetic test
- Whether state programs should implement parental permission for newborn screening
- Implementation challenges such as modifying health records to include genetic data and training health professionals on the uses and limitations of new technologies
- Distinctions between genetic tests that provide information about the health of the person being tested and those that identify the person as a potential carrier of disease
- Ways in which children of different ages understand and act upon genetic findings
- Questions related to adoption, consanguinity, and paternity

The advent of WGS raises questions about how extensively it should be used. At this time, the statement authors did not recommend routinely sequencing the genomes of healthy children, including newborns. In contrast, they said, for a child likely to have a genetic disease, WGS would be an appropriate diagnostic tool, but the scope of testing should be restricted to a single gene or limited set of genes when possible.

The statement authors also outlined areas in which additional research is needed. An important challenge is the gap between scientists' ability to identify genetic variants and their ability to interpret what they mean. For this reason, the authors recommend that geneticists develop a list of genes for which the chromosomal variations detected by CMA are clearly related to disease. Along similar lines, they called for further evaluation of pharmacogenetic testing to assess its usefulness and limitations in predicting how children respond to drugs.

In addition, the authors stated, adequate data on the psychosocial impact of pediatric genetic testing (including carrier testing) on children and their families is lacking. Clear communication of results is key, they noted, and recommended additional research into the best ways to educate children and parents before the test and to explain results after the test.

"Genetic testing provides powerful information about an individual's future and that of their family, which enables informed decision making about healthcare, lifestyle, and reproduction - particularly impactful for children and adolescents who are early in their lives. At the same time, this information is largely probabilistic, making it difficult to communicate clearly to patients and their parents, and it may trigger stigma or discrimination," said Joseph D. McInerney, MS, Executive Vice President of ASHG.

"For these reasons," Mr. McInerney added, "it is especially important to carefully consider the implications of genetic testing in children and adolescents and choose the best way to implement these new technologies."

Provided by American Society of Human Genetics