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***Objective**: To promote dialogue among readers and between readers and editors, we offer here brief reports and observations on topics of interest authored by members of the AAP Grand Rounds editorial team.*

New Guidelines on Genetic Testing and Screening in Children

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Dr. Hamid has disclosed no financial relationship relevant to this commentary. This commentary does not contain a discussion of an unapproved/investigative use of a commericial product/device.

Recent advances in genetics and genomics have led to a tremendous increase in the availability and use of genetic testing and screening in children, including genetic assays marketed directly to consumers. Accompanying this trend are significant ethical issues. The American Academy of Pediatrics (AAP) and the American College of Medical Genetics and Genomics (ACMG) recently updated their policy statements regarding genetic testing in children to inform best practices.[1,2] These are comprehensive guidelines that address many commonly encountered clinical scenarios. A summary of the major guideline recommendations are provided below:

1. Newborn screening: The AAP and ACMG support the mandatory offering of newborn screening for all children but advise that parental permission be sought. In 2002, the ACMG recommended universal screening for a panel of 29 primary targets, which has since been endorsed by the US Department of Health and Human Services and adopted by all states.

2. Diagnostic genetic testing: Genetic testing for children with symptoms of a genetic condition is akin to any other medical diagnostic test. Parental permission should be obtained and the benefits and harms of the test considered (including discovery of misattributed parentage). When the medical benefit-burden ratio of a genetic test is unfavorable or uncertain, or the benefits won't accrue until a later time, there is less justification for performing the genetic test.

3. Carrier testing: The AAP and ACMG do not recommend routine carrier testing of children for autosomal recessive disorders except in the circumstances when carrier status has potential medical implications during childhood. For example, carrier screening may be appropriate for adolescents who are pregnant.

4. Predictive genetic testing: The AAP and ACMG recommend deferring predictive genetic testing for late-onset disorders until adulthood. However, predictive genetic testing may be appropriate in limited circumstances but ought to be guided by the child's best interests.

5. Direct-to-consumer genetic testing: The AAP and ACMG strongly discourage the use of this type of genetic testing in children given concerns about inaccurate results and unreliable interpretation.

6. Disclosure of genetic test results: The AAP and ACMG recommend giving priority to mature adolescents' requests for genetic test results over requests by parents to conceal this information. For nondisclosure requests by parents of young children, the provider should work with the parents to develop a plan for disclosing the existence of the test (and its results) when the child reaches adulthood.

References

1. Ross LF, et al. *Genet Med.* 2013;15(3):234-245; doi:10.1038/gim.2012.176
2. Committee on Bioethics. *Pediatrics.* 2013;131(3):620-622; doi:10.1542/peds.2012-3680

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