Genetic testing in kids is fraught with complications

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American Society of Human Genetics (ASHG) Workgroup on Pediatric Genetic and Genomic Testing has issued guidelines for genetic testing in children and adolescents that are based on a thorough review of studies on ethical, legal, and social implications (ELSI). The recommendations were published in the American Journal of Human Genetics. The workgroup is chaired by bioethicist Jeffrey Botkin, M.D., M.P.H., director of the Utah Center for Excellence in Ethical Legal and Social Implications Research (UCEER) at the University of Utah School of Medicine. Credit: University of Utah Health Sciences

A woman coping with the burden of familial breast cancer can't help but wonder if her young daughter will suffer the same fate. Has she inherited the same disease-causing mutation? Is it best to be prepared for

the future, or to wait?

During the last decade, genetic tests have been through a sea change, both in their availability and the technologies behind them. Today there are at least 34 companies that offer direct to consumer (DTC) DNA testing, some of which return health results. And now it is possible to sequence someone's entire genetic code for the price of a laptop.

Despite an increasing ease in acquiring genetic information, the American Society of Human Genetics (ASHG) points out that doing so has consequences, particularly when it comes to children. It is this population, they say, that is the most vulnerable.

With this precaution in mind, the ASHG Workgroup on Pediatric Genetic and Genomic Testing has issued guidelines for genetic testing in children and adolescents that are based on a thorough review of studies on ethical, legal, and social implications (ELSI). The recommendations were published in *The American Journal of Human Genetics*.

"Kids are always considered to be a vulnerable population because they can't make decisions for themselves," says bioethicist Jeffrey Botkin, M.D., M.P.H., chair of the workgroup and director of the Utah Center for Excellence in ELSI Research (UCEER) at the University of Utah School of Medicine. "It's important to be respectful of the parents' right to make decisions for them, but at the same time the situation can lead to challenging circumstances."

Bioethicists agree that genetic testing in children is nearly always warranted when the results could have an immediate impact on health care decisions. For example, if searching for the cause of a serious illness, or if a child is at imminent risk for developing a disease such as childhood cancer.

It is predictive testing - looking for genetic signs of health conditions that typically arise during adulthood - that is particularly fraught with complication.

When a Child is Young, It May Be Best to Wait

Take the example of the mother with breast cancer. She may decide to have her six-year-old daughter tested because she presumes it's best for the girl and their family. But once the girl grows up, she may see things differently.

"It is acceptable for a 25-year-old to decide not to have knowledge about what is likely to happen to her when she's 60. Following that rationale, a parent should not be able to make a decision for a six-year-old girl that would prevent her from making a different decision at a later age," says Benjamin Wilfond, M.D., a physician and bioethicist at Seattle Children's Hospital. He is also a member of the working group and was lead author of ASHG's first set of guidelines issued twenty years ago.

Further, the results could affect the relationship between the girl and her parents. "Physicians and bioethicists have been concerned that this powerfully predictive information could stigmatize the child. We all hope for bright futures for our children but a genetic prediction might create a dark cloud that changes how the family and others think about the prospects of that young girl for health, but also for a career and marriage," explains Botkin.

The stress may not only be challenging for the family, but it could also be unfounded. The vast majority of genetic tests don't outright predict someone's health future. Sometimes the disease never develops, even if it's against the odds. Based on considerations like these, ASHG recommends holding off on this type of predictive testing until adulthood when the child can make this decision herself.

Acknowledging Adolescents

But, with every rule comes exceptions. For most, studies show, the uncertainty of not knowing is a greater psychological burden than living with bad news.

Flash forward ten years and the girl, now 16, is old enough to understand why breast cancer runs in her family. She's anxious about her own future and wants to know whether she carries the mutation.

"There may be individual circumstances where testing during childhood is an acceptable choice," says Botkin. In situations like this one, it could be best for the teen to talk to a trusted health care professional - without her parents - for an unbiased assessment of the benefits and risks of testing. Ideally, the teen and parents would come to a final decision together.

Under any circumstance, ASHG recommends against DTC genetic testing, especially for children. At this point in time, the tests are too unreliable, and results are usually returned without sufficient interpretation or guidance. "Kids are better served by testing within the health care setting, where they get proper counseling and decision making support," says Botkin.

Target the Trouble

Whole-genome sequencing (WGS) comes with the promise of accessing all the health information encoded within our DNA. While useful in a number of clinical situations when targeting testing is not helpful, ASHG recommends against doing so in healthy children.

Among the potential problems is finding out too much too soon. What if

the test reveals an unanticipated risk for another adult onset disease? The scenario stirs up the same concerns as those experienced by the girl and her family. Not to mention that because the health risk is by definition inherited, it may have unwelcome implications for parents, brothers and sisters, or more distant relatives as well.

As a result, ASHG recommends only testing for the genes(s) expected to be involved with health issues of immediate concern. Or if WGS is performed, limiting analysis to the genes of interest is ethically acceptable.

The published guidelines cover several additional issues including paternity, newborn screening, and pharmacogenomics testing.

"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," says Joseph D. McInerney, M.S., Executive Vice President of ASHG. "For these reasons 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 University of Utah Health Sciences