# CVD genetic testing in children presents unique challenges, needs individualized approach

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Cardiovascular genetic testing in children presents unique challenges, requiring pre- and post-test counseling with an individualized approach for families, ideally with the involvement of a specialized interdisciplinary team, according to a new American Heart Association scientific statement published today in the Association's journal *Circulation: Genomic and Precision Medicine*. Scientific statements represent the synthesis of data and a consensus of the leading experts, designed to address gaps in guidelines.

This is the first American Heart Association scientific statement providing guidance specifically about genetic testing of cardiovascular diseases in children. In addition to previous statements issued by the Association on genetic testing mostly focused on adults, it was important to issue a pediatric-focused statement with the recognition that children and their families face unique challenges specific to pediatric gene testing, according to the statement writing group.

"There is growing recognition that a genetic test is not a simple blood test where you get a yes or no answer. With this statement, we illustrate some of that complexity, particularly as it relates to cardiovascular diseases passed from parents to children," said Andrew P. Landstrom, M.D., Ph.D., FAHA, chair of the statement writing group, a pediatric cardiologist, a cardiovascular geneticist and an assistant professor of pediatrics and cell biology at Duke University School of Medicine in Durham, North Carolina. "We provide consensus-based recommendations for best practices and principles to assist health care professionals in determining when cardiovascular gene testing is

appropriate in children, highlighting the need for a multi-disciplinary approach to family counseling before and after testing, and we raise the importance of appropriate follow-up."

The statement writing group noted that pre-test counseling is essential in genetic testing of children. Before a decision is made about genetic testing, counseling should be held with the parents and with the child, if the child is old enough to fully comprehend and able to contribute to the decisions. Counseling should cover the possible benefits of genetic testing and the limits of the test's ability to help with diagnosis and management, along with the possible outcomes of testing, including the potential impact on care. Pre-test counseling should address the possibility that the genetic results may be inconclusive since there is still much to be discovered about the genetic components of many diseases of the heart. Clinicians must collaborate with the family to be prepared for all scenarios, before testing is conducted—including a positive genetic test, a negative test or whether the test is inconclusive.

Pre-test counseling is also the time to address family concerns about possible medical costs or the possibility that genetic test results could lead to discrimination or an inability to obtain health insurance in the future. The statement details federal laws families should know about that can help to alleviate some of these concerns.

Post-test counseling and follow-up are also essential, according to the statement. After genetic test results are in, post-test counseling can be a time to explain the findings and plan how the information can be used in caring for the person affected, as well as proceeding with testing or treatment for other members of the family. Ongoing follow-up, possibly over the course of a lifetime, is also important because, in the rapidly evolving field of genetics, the understanding of a specific gene's significance may change over time.

"It's important to understand what's ahead before you start the process. In a disease such as long QT syndrome (a disorder of the heart's electrical system) or catecholaminergic polymorphic ventricular tachycardia (an inherited arrhythmia) genetic testing can identify a likely cause of the disease 60%-75% of the time. This means if it's suspected that an individual has one of these diseases, the test is likely to find the gene variant that causes the disease about two-thirds of the time. Identification of the presence or absence of this gene variant in family members of the individual can identify those who are, or are not, at risk of developing the same disease. In this situation, gene testing makes sense in most scenarios," Landstrom said. "With some inherited cardiovascular diseases, the genetic causes are not well known and the likelihood of finding the gene causing the condition is much lower in pediatric testing. For these diseases, the likelihood of finding one of these genetic variants of uncertain significance can make the test challenging to interpret. These are important considerations for clinicians and families to understand and discuss together before testing is done."

The writing group advises that as genetic testing becomes more readily available, it's important that it be used appropriately. Clinicians should first determine or have a strong sense of a clinical diagnosis—the genetic testing can help refine the diagnosis and, in some cases, can help define the next of many possible steps in disease management.

Two main types of cardiovascular gene testing are provided to children—diagnostic and risk-predicting.

If, after a thorough cardiology workup, a child is strongly suspected of having a hereditary heart condition, a diagnostic genetic test may be ordered to determine whether the child has the gene variation known to be associated with that condition. A diagnostic gene test can refine the clinical diagnosis and help with decisions about how best to manage the

condition in some cases, such as choosing a medication or recommending lifestyle changes that can improve the likelihood of the person staying healthy.

The second type of testing, called risk-predictive testing, is done on a close relative, such as a sibling, parent or a child of the person found to have a genetic variant associated with a heart condition.

"If a relative is found to have the same gene variant that was determined to be the likely cause of disease in an affected family member, the relative may be at-risk for developing the same condition. This doesn't mean they are guaranteed to develop the condition in the same way, or even that they will get the condition at all, just that they are 'at-risk', and they will need to be closely monitored because of this increased risk," Landstrom said. "Conversely, if the gene variant causing the condition in the family is identified and a relative tested doesn't have that genetic variant, that person likely does not need additional follow-up or screening beyond that of the general population."

The statement also addresses gene-sequencing, another type of gene testing gaining momentum in recent years, which involves testing children in whom there is no suspicion for cardiac disease but may alternatively identify another genetic syndrome. As genetic sequencing has gotten easier and less expensive, it is more available to clinicians, and more of it is being done. That convergence of technology and clinical availability of genetic tests makes it important to provide guidance to be clear on the rationale for ordering a genetic test and ensuring tests are not ordered inappropriately or just because they are available.

Prior to initiating genetic testing, practitioners should consider referring a child with a suspected heritable cardiovascular disease to a multidisciplinary cardiovascular genetics program. Multidisciplinary

teams typically include close collaboration among adult and pediatric cardiologists, genetic counselors, geneticists, behavioral health specialists and others as appropriate for a case. While these programs are usually found at large medical centers, improved telehealth capability has made access easier.

"Pediatric genetic testing has important considerations beyond those of adult testing, including the vulnerability of children as a population, and these considerations should be at the forefront of all decision-making about genetic testing," Landstrom said. "Other considerations include the dynamics within the child's family, the family's goals and concerns, potential psychosocial effects of testing (or not testing) and the current state of genetic testing methods. Additional factors to help inform the decision-making process to ensure the best outcomes for children and family members are the characteristics of the specific heritable cardiovascular disease, the likelihood and timing of disease development, the availability of therapies and interventions that can treat or prevent disease, and the availability and cost of testing."

**More information:** Andrew P. Landstrom et al, Genetic Testing for Heritable Cardiovascular Diseases in Pediatric Patients: A Scientific Statement From the American Heart Association, *Circulation: Genomic and Precision Medicine* (2021). DOI: 10.1161/HCG.0000000000000086

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