Medical Xpress

Growing cardiovascular genetics field calls for special multidisciplinary clinical programs

May 23 2019

With a better understanding of how various heart conditions are inherited, and the availability of faster and less expensive genetic testing, there is need for more specialized multidisciplinary clinical programs that combine focused expertise in heart disease and genetics, according to a new statement from the American Heart Association, the world's leading voluntary organization focused on heart and brain health. The statement is published in the Association's journal *Circulation: Genomic and Precision Medicine*.

Cardiovascular genetics, as a subspecialty, has grown exponentially with the advances in genome sequencing and genetic testing and the growing understanding of the genetic basis of multiple heart conditions. Challenges exist with rapid growth, including the interpretation of genetic test results and the evaluation, counseling and management of genetically at-risk family members who have inherited the genetic alteration that increases their predisposition to a certain disease even if they have not yet shown signs or symptoms.

"The state of genetic understanding of some heart disorders has improved to the point that we can use this information to help families and offer hope in ways never before possible," said Ferhaan Ahmad, M.D., Ph.D., chair of the writing group for the statement, associate professor of cardiovascular medicine and molecular physiology and director of the cardiovascular genetics program with University of Iowa

Health Care in Iowa City. "But it's important that we have the right people, including medical geneticists and genetic counselors, as well as adequate facilities, equipment and other resources in place to provide clear and accurate guidance to these families throughout testing and decision-making processes."

The statement lays out what would be needed to create a quality specialized program:

- Leadership from a cardiologist well-versed in genetics or a geneticist well-versed in cardiovascular medicine;
- Core personnel including cardiologists, medical geneticists, genetic counselors, nurse managers and clinic coordinators;
- Facilities for several types of cardiac imaging and people with the expertise to interpret the findings and recognize uncommon heart conditions;
- The subspecialists and facilities to offer (either at the program or with a well-developed referral plan for complex surgeries) the invasive procedures necessary to diagnose and treat electrical problems in the heart, to assess heart function, to surgically correct structural problems of the heart, valves and aorta and to perform heart transplants;
- Genetic testing and counseling; and
- Support from other specialties such as sleep medicine, behavioral medicine, nutrition, social work and exercise physiology.

Currently, there are a few broad-based cardiovascular genetics programs in existence at academic centers, as well as smaller programs focused on a specific disease, such as hypertrophic cardiomyopathy—the most common genetic disorder of the heart, which results in a thickened heart muscle that has a harder time pumping blood.

Statement authors said a specialized genetic program that provides the

integration of clinical cardiovascular findings—including those obtained from physical examination, imaging and functional assessment—with genetic information allows for improved diagnosis, prognosis and generational family testing to identify and manage risk and in certain cases to provide genotype-specific therapy.

Specialized programs in cardiovascular genetics could benefit both those with inherited heart conditions and their healthy family members, according to the statement.

"A patient who has a rare, genetic heart disease will benefit from specialized care from experts who know how to manage a disease that's not familiar to the general cardiologist," said Kiran Musunuru, M.D., Ph.D., M.P.H, associate professor of cardiovascular medicine and genetics at the University of Pennsylvania and outgoing editor-in-chief of *Circulation: Genomic and Precision Medicine*, who wasn't involved in the development of the guideline. "Others having a close relative with an inherited heart disease can be screened to see if they have inherited genes that put them at risk for getting the disease in the future. If so, they can be monitored over time for early signs of disease and they can, in some cases, be treated to prevent the most serious consequences of the disease."

Ahmad said in addition to providing quality care to families, these centers will also become key places for the genetics training of internal medicine and pediatric residents and cardiology fellows.

The American Heart Association is prepared to be part of the educational efforts needed to translate advances in genetics into better care for families with heart disease.

"The American Heart Association, led by the Council on Genomic and Precision Medicine and the Institute for Precision Cardiovascular

Medicine, is primed and ready to support the education of clinicians and researchers in genetics and data science through the Precision Medicine Platform," said Jennifer L. Hall, Ph.D., Chief of the American Heart Association's Institute for Precision Cardiovascular Medicine. "We're connecting clinicians and researchers by funding innovative data science, opening up new data sources with rich genetic information and allowing collaborators to analyze data in real time together on the Precision Medicine Platform in secure workspaces equipped with high performance computing and analytical tools."

Other types of genetic diseases that might be diagnosed and treated at a specialized program include those that result in abnormally high levels of bad cholesterol, vascular disorders such as Marfan syndrome that can weaken the aorta, and abnormalities of heart rhythm that can raise the risk of sudden death. While cardiovascular genetics programs may serve both adults and children, these programs and the new scientific statement are unrelated to the treatment of congenital heart defects—structural birth defects of the heart that are not inherited.

More information: *Circulation: Genomic and Precision Medicine*, DOI: 10.1161/HCG.0000000000000054

Provided by American Heart Association