

EVALUATING PATIENTS WITH A CONFIRMED OR SUSPECTED DIAGNOSIS OF INHERITED CARDIOVASCULAR DISEASE

According to the American Heart Association, all cardiovascular practitioners should have the ability to evaluate whether their patients might have genetic cardiovascular conditions.

Below are the key components that all cardiovascular practitioners should consider when evaluating patients with a confirmed or suspected diagnosis of an inherited cardiovascular disease.



Practitioners must consider two elements when identifying patients for genetic testing:

- ◆ Disease-appropriate phenotyping to establish suspected diagnosis
- ◆ Work with the patient to document their family medical history going back three generations to determine if there is a pattern of certain types of heart disease.



If genetic testing is deemed appropriate, the clinician should decide what scope of genetic testing needs to be performed and identify the appropriate patient(s) for testing (patient or family member)

The choice of testing ranges from targeted sequencing of a single gene or a few genes most likely to be involved in the disease, to large gene panels that include limited-evidence genes, to unbiased exome or genome sequencing that queries all genes.



Genetic counseling is essential before genetic testing to educate patients on the process and potential results, as well as the potential risks and uncertainties related to testing. Counseling is also critical after genetic testing, so the counselor can explain the results and potential consequences for the patient's health and the health of family members.

The finding of a pathogenic or likely pathogenic diagnostic variant should prompt an attempt to perform cascade genetic testing of family members.



Reference: Musunuru, K., Hershberger, R. E., Day, S. M., Klinedinst, N. J., Landstrom, A. P., Parikh, V. N., Prakash, S., Semsarian, C., Sturm, A. C., & American Heart Association Council on Genomic and Precision Medicine; Council on Arteriosclerosis, Thrombosis and Vascular Biology; Council on Cardiovascular and Stroke Nursing; and Council on Clinical Cardiology. (2020). Genetic Testing for Inherited Cardiovascular Diseases: A Scientific Statement From the American Heart Association. *Circulation. Genomic and Precision Medicine*, 13(4), e000067.

Want to learn more about genomic testing in cardiology?

Visit our website: WWW.CARDIOGENOMICTESTING.COM

