REFERENCE GUIDE

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A Practical Guide to Cardiovascular Genetic Testing and Selection in Clinical Care

Without genetic insight, high-risk cardiovascular patients may be overlooked.

A major study found that although **1.7%** of patients carried a monogenic cardiovascular condition, **65%** were completely undiagnosed, underscoring the critical role of genetic testing in identifying hidden risk.[1] By identifying the molecular etiology of disease, genetic testing can improve diagnostic accuracy and refine family management.[2]

The case for cardiovascular genetic testing in your practice:

- **Diagnostic clarification:** Clarify or confirm the underlying patient diagnosis[2]
- **Informed decision-making:** Guide personalized, evidence-based management recommendations
- **Risk stratification and management:** Reduce the risk of genotype-specific cardiac events
- **Early detection:** Enable proactive screening and prevention for at-risk relatives[2]
- **Align to guidelines:** Ensure your practice standards and protocols are keeping current with guideline-recommended genetic testing

Despite guideline recommendations, genetic testing is underutilized.[3]

Apply guideline-recommended genetic testing with confidence

Genetic testing is supported across major cardiology and genetics societies for key inherited cardiovascular conditions. Societies like the American Heart Association (AHA), American College of Cardiology (ACC), the Heart Rhythm Society (HRS), the European Heart Rhythm Association (EHRA), and the Heart Failure Society of America (HFSA) provide guidance to help you determine when genetic testing is appropriate.

Condition	AHA/ACC[4,5]	HRS/EHRA[6–8]	HFSA[9]	Consensus
Hypertrophic cardiomyopathy (HCM)	●	●	●	●[10]
Dilated cardiomyopathy (DCM)	●	●	●	●[11]
Arrhythmogenic right ventricular cardiomyopathy (ARVC/ACM)	●	●	●	
Long QT syndrome (LQTS)/Brugada syndrome (BrS)	●	●		●[12]
Catecholaminergic polymorphic ventricular tachycardia (CPVT)	●	●		
Familial thoracic aortic aneurysm and dissection (TAAD)	●			●[13]
Familial hypercholesterolemia (FH)	●			●[14]
Sudden unexplained death	●	●		
Amyloidosis	●			●[15]

Key indicators for cardiovascular genetic testing:

Patients with a personal or family history of any of these clinical features should be considered for genetic testing. The best candidate for genetic testing is typically those family members affected with the cardiovascular condition. However, unaffected or asymptomatic people may be tested when an affected relative is unavailable. Consider genetic testing when there is a/an:

- Clinical diagnosis of ARVC/ACM, DCM, HCM, LQTS, BrS, CPVT, TAAD, FH
- Unexplained arrhythmia
- Sudden cardiac arrest or death (SCA, SCD)
- Recurrent fainting spells
- Early-onset heart disease (<55 yo in men, and <65 yo in women)
- Heart failure or heart transplant
- Presence of a pacemaker or implantable cardioverter-defibrillator (ICD), especially at a young age
- Suspicion of syndromic cardiovascular condition (e.g., Noonan syndrome, Marfan syndrome)
- Multiple affected family members across generations

Select the right cardiovascular genetic test

Choose from an extensive menu of cardiovascular genetic testing options designed to clarify inherited risk and guide clinical action. Targeted panels (4–50 gene panels) focus on specific cardiovascular conditions such as cardiomyopathy, arrhythmia, aortopathy, and lipid disorders. Or, select a comprehensive panel such as CardioNext,® (92 genes) which includes genes associated with both cardiomyopathies and arrhythmias .

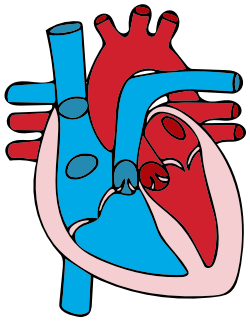
Aortic or Vascular testing includes:

Targeted Panels
- TAADNext® (35 genes)
- HHTNext® (6 genes)

Arrhythmia testing includes:

Targeted Panels
- CPVTNext® (4 genes)
- LongQTNext™ (17 genes)

Multi-condition Panels
- RhythmNext® (42 genes)
- CardioNext® (92 genes)



Lipid disorder testing includes:

Targeted Panels
- FHNext® (4 genes)
- FCSNext® (5 genes)
- Sitosterolemia (2 genes)

Cardiomyopathy testing includes:

Targeted Panels
- HCMNext® (30 genes)
- DCMNext® (37 genes)
- ARVCNext™ (11 genes)

Multi-condition Panels
- CMNext® (56 genes)
- CardioNext® (92 genes)

Postmortem testing includes:

Testing options
- Select appropriate panel based on autopsy findings

Other conditions testing includes:

Targeted Panels
- NoonanNext™ (18 genes)

What to know before ordering

Expertise that matters

We maintain the highest commitment to quality genetic testing. Our clinical team uses advanced research tools, collaborates with partner labs, and identifies updates that may impact variant classifications. We also have 100+ genetic counselors and other medical experts who can support result interpretation, discuss complex cases, and ensure you receive clear, accurate guidance.

Flexible sample options

We accept blood, buccal swab, saliva, and other sample types to make it easier for you to collect the right specimen in any clinical setting. This flexibility helps reduce barriers to testing and supports faster, more convenient ordering.

Coverage, pricing, and patient support

Many commercial and Medicaid plans now recognize cardiogenetic testing, and Ambry can help streamline insurance verification and prior authorization. When needed, patients can access transparent self-pay pricing and financial assistance. Ask your Ambry representative for billing resources or contact our billing support team at +1 949-900-5500.

Access to third-party genetic counseling services

We recognize that not all organizations have access to a local genetics expert. We provide access to third-party genetic counselors who can support your practice and your patients.

For support selecting the right panel for your patient, connect with your Ambry Genetics account executive, visit www.ambrygen.com/cardio or reach us at +1 949-900-5500.

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