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Recommendations & Guidelines for Cardiovascular Genetic Testing

GENETIC TESTING IS RECOMMENDED/BENEFICIAL *(includes Class I recommendations)*	AHA/ACC	HRS/EHRA	HFSA	CONSENSUS / PANELS
Hypertrophic Cardiomyopathy (HCM)	✓5, 11	✓10	✓2	✓1
Dilated Cardiomyopathy (DCM)	✓5	✓10	✓2	✓3
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)	✓5	✓10, 8	✓2	
Arrhythmic disorders / Long QT (LQTS), Brugada Syndrome (BrS), Type 1	✓5	✓10		✓4
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)	✓5	✓10		
Thoracic Aortic Aneurysms and Dissections	✓5			✓9
Familial Hypercholesterolemia	✓5			✓7
Sudden unexplained death and sudden cardiac arrest	✓5,11	✓10, 6		

AHA = American Heart Association, ACC = American College of Cardiology, HRS = Heart Rhythm Society, EHRA = European Heart Rhythm Association, HFSA = Heart Failure Society of America

1. Cirino AL, Ho C. Hypertrophic Cardiomyopathy Overview. 2008 Aug 5 [updated 2019 Jun 6]. In: Adam MP, et al. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2020.
2. Hershberger RE, et al. Genetic Evaluation of Cardiomyopathy-A Heart Failure Society of America Practice Guideline. *J Card Fail.* 2018 May;24(5):281-302.
3. Hershberger RE, Morales A. Dilated Cardiomyopathy Overview. 2007 Jul 27 [updated 2018 Aug 23]. In: Adam MP, et al. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2020.
4. McNally E, et al. Arrhythmogenic Right Ventricular Cardiomyopathy. 2005 Apr 18 [updated 2017 May 25]. In: Adam MP, et al. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2020.
5. Musunuru K, et al. Genetic Testing for Inherited Cardiovascular Diseases: A Scientific Statement From the American Heart Association. *Circ Genom Precis Med.* 2020 Aug;13(4):e000067.
6. Stiles MK, et al. 2020 APHRS/HRS expert consensus statement on the investigation of decedents with sudden unexplained death and patients with sudden cardiac arrest, and of their families. *Heart Rhythm.* 2021 Jan;18(1):e1-e50.
7. Sturm AC, et al. Clinical Genetic Testing for Familial Hypercholesterolemia: JACC Scientific Expert Panel. *J Am Coll Cardiol.* 2018 Aug 7;72(6):662-680.
8. Towbin JA, et al. 2019 HRS expert consensus statement on evaluation, risk stratification, and management of arrhythmogenic cardiomyopathy. *Heart Rhythm.* 2019 Nov;16(11):e301-e372.
9. Verhagen JMA, et al. Expert consensus recommendations on the cardiogenetic care for patients with thoracic aortic disease and their first-degree relatives. *Int J Cardiol.* 2018 May 1;258:243-248.
10. Wilde AAM, et al. European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the state of genetic testing for cardiac diseases. *Europace.* 2022 Apr 4:euac030.
11. Writing Committee Members et al. 2020 AHA/ACC Guideline for the Diagnosis and Treatment of Patients With Hypertrophic Cardiomyopathy: A Report of the American College of Cardiology/American Heart Association Joint Committee on Clinical Practice Guidelines. *Circulation.* 2020 Dec 22;142(25):e558-e631.

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Best Practices for Cardiovascular Genetic Testing

BEST PRACTICE	HOW AMBRY HELPS
Patient Identification – In patients with HCM, DCM, arrhythmic disorders, thoracic aorta disease, or dyslipidemia – a comprehensive 3 generation family history should be obtained	Genomic Science Liaisons (GSL) available to assist with patient identification strategies
Genetic Counseling – Genetic testing is most effective when accompanied by pre and/or post-test genetic counseling	Genetic counseling options available for your patients
Multigene Panel Test – Genes should include those with strong clinical evidence	Highly curated panels with moderate or higher clinically valid genes
Genetic Variants – Routine reevaluation of identified genetic variants is recommended for variant reclassification (could impact diagnosis and family testing)	On-going evaluation of all variants within the lab; amended reports issued with changes in classification
Cascade Testing – Should be offered to all at-risk relatives of genotype-positive proband to facilitate identification of family members at risk for cardiac disease	No-cost cascade testing for 90 days from date of proband report
Family Member Screening – Individuals who are genotype-positive should have regular clinical surveillance	GSL team can review condition-specific recommendations
Postmortem Testing – Genetic testing in cases of sudden unexplained death can be beneficial in identifying family members for screening	Postmortem team facilitates specimen logistics; cascade testing for at-risk family members

"Counseling patients with HCM regarding the potential for genetic transmission of HCM is one of the cornerstones of care."

AHA, 2020[11]

"Genetic testing is informative and useful for the clinical management of various inherited cardiovascular diseases such as cardiomyopathies, arrhythmic disorders, thoracic aortic aneurysms and dissections, and familial hypercholesterolemia (FH)."

AHA, 2020[5]

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