

Cardiogenetics Testing

Reference Guide

Why Choose Ambry

Approximately 1 in 60 people have a hereditary cardiovascular condition.^{1,2} Ambry's mission is to provide the most advanced genetic testing information available to help you identify those at-risk and determine the best treatment options. If we know a patient has a disease-causing genetic change, not only does it mean better disease management, it also indicates that we can test others in the family and provide them with potentially life-saving information.

Diseases and Testing Options

ARRHYTHMIAS

Catecholaminergic Polymorphic Ventricular Tachycardia (**CPVTNext®**)

Long QT Syndrome, Short QT Syndrome, Brugada Syndrome (**LongQTNext, RhythmNext®**)

Arrhythmias (**RhythmNext, CardioNext®**)

CARDIOMYOPATHIES

Hypertrophic Cardiomyopathy (**HCMNext®**)

Dilated Cardiomyopathy (**DCMNext®**)*

Arrhythmogenic Right Ventricular Cardiomyopathy (**ARVCNext**)

Cardiomyopathies (**CMNext®, CardioNext**)

OTHER CARDIO CONDITIONS

Hereditary Transthyretin Amyloidosis (**TTR**)

Noonan Syndrome (**NoonanNext**)

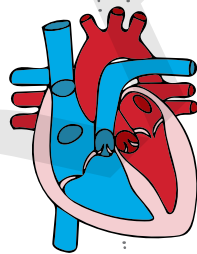
Hereditary Hemorrhagic Telangiectasia (**HHTNext®**)

THORACIC AORTIC ANEURYSMS AND DISSECTIONS

Thoracic Aneurysms and Dissections, aortopathies (**TAADNext®**)

Marfan Syndrome (**TAADNext**)

Ehlers-Danlos Syndrome (**TAADNext**)



FAMILIAL HYPERCHOLESTEROLEMIA AND LIPID DISORDERS

Familial Hypercholesterolemia (**FHNext®**)

Sitosterolemia (**Sitosterolemia Panel**)

Familial Chylomicronemia Syndrome (**FCSNext**)

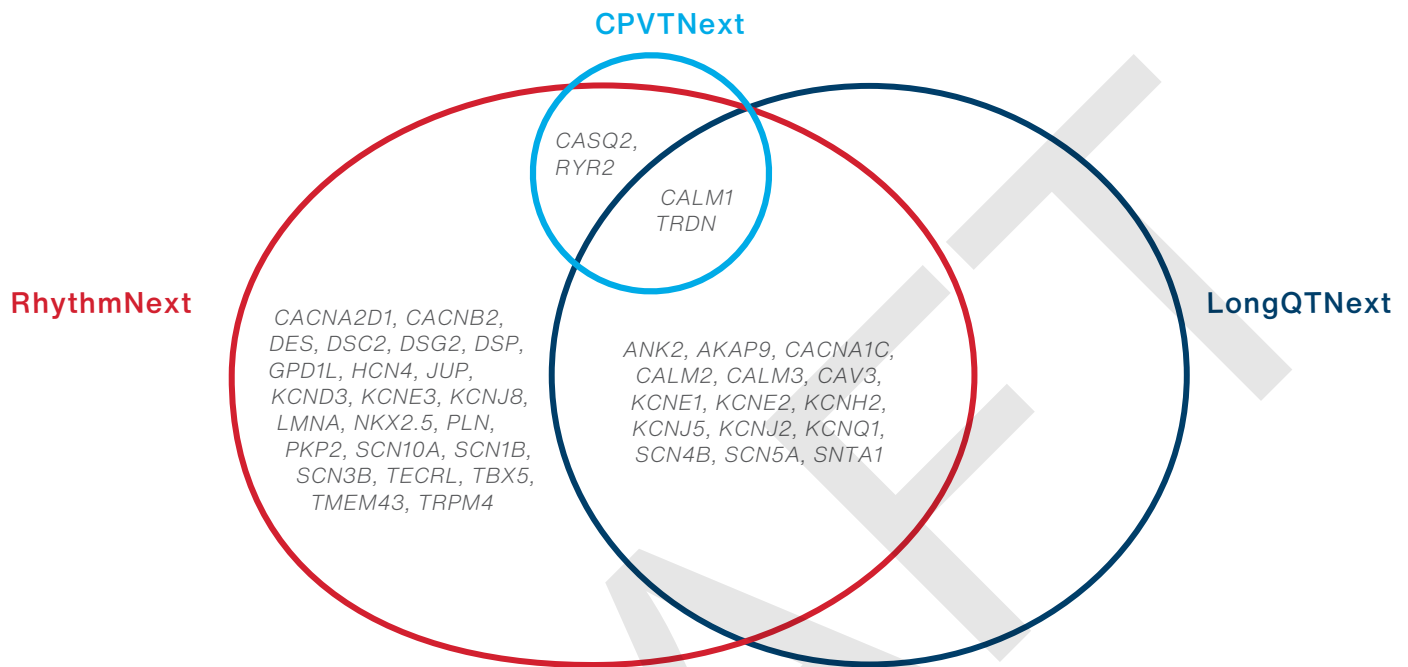
Comprehensive Lipid Menu (**CustomNext-Cardio®**)

* Genes associated with Left Ventricular Non-Compaction (LVNC) are included in DCMNext.

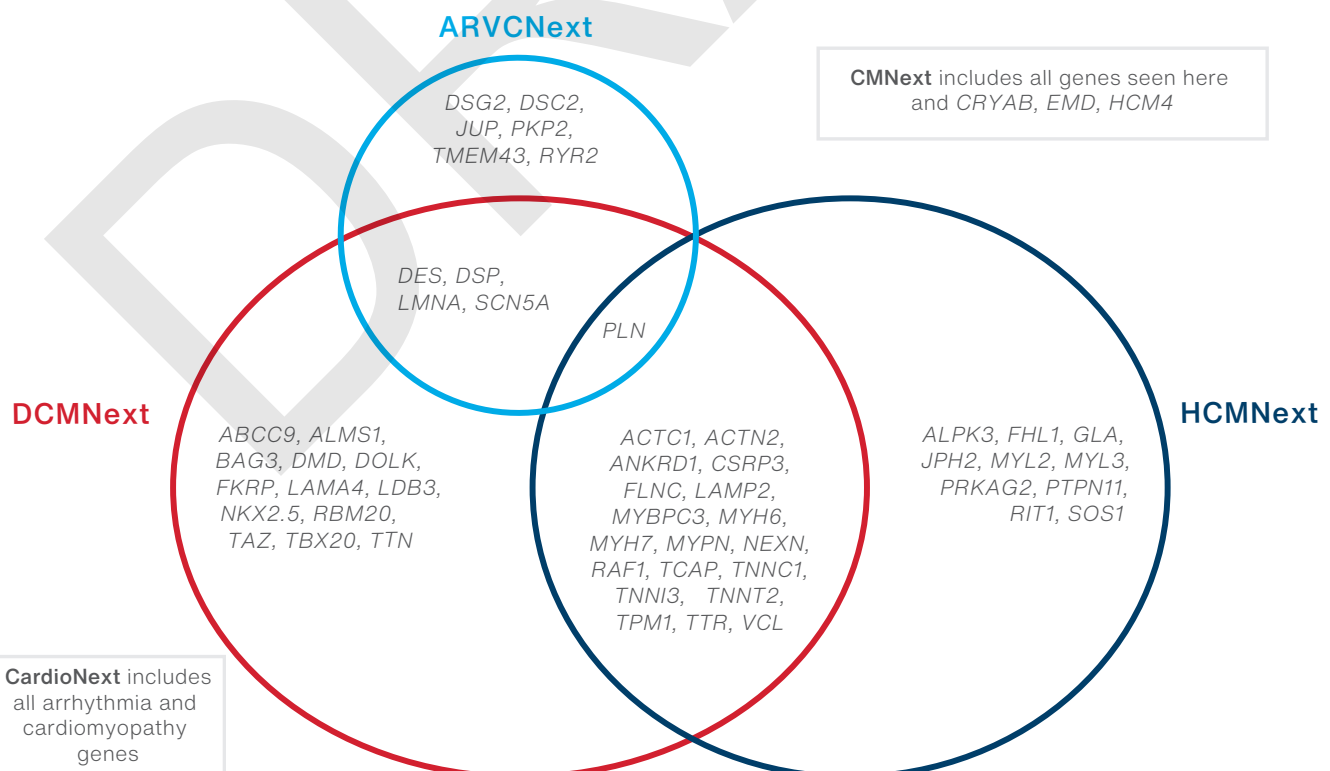
Targeted Panels Gene Comparison

All panels have a turnaround time of 2-3 weeks

ARRHYTHMIAS



CARDIOMYOPATHIES



Comprehensive, Targeted, and Custom Panels Gene Lists

All panels have a turnaround time of 2-3 weeks

CardioNext 92 genes		TAADNext 35 genes	NoonanNext 18 genes	FHNext 4 genes + SNP
ABCC9	LAMA4	ACTA2	BRAF	APOB
ACTC1	LAMP2	BGN	CBL	LDLR
ACTN2	LDB3	CBS	HRAS	LDLRAP1
AKAP9	LMNA	CHST14	KRAS	PCSK9
ALMS1	MYBPC3	COL1A1	LZTR1	SLCO1B1 (c.521T>C)*
ALPK3	MYH6	COL1A2	MAP2K1	
ANK2	MYH7	COL3A1	MAP2K2	
ANKRD1	MYL2	COL5A1	NF1	
BAG3	MYL3	COL5A2	NRAS	
CACNA1C	MYOZ2	EFEMP2	PPP1CB	
CACNA2D1	MYPN	FBN1	PTPN11	
CACNB2	NEXN	FBN2	RAF1	
CALM1	NKX2.5	FKBP14	RASA1	
CALM2	PKP2	FLNA	RIT1	
CALM3	PLN	FOXE3	SHOC2	
CASQ2	PRKAG2	LOX	SOS1	
CAV3	PTPN11	MAT2A	SOS2	
CRYAB	RAF1	MED12	SPRED1	
CSRP3	RBM20	MFAP5		
DES	RIT1	MYH11		
DMD	RYR2	MYLK		
DOLK	SCN10A	NOTCH1		
DSC2	SCN1B	PLOD1		
DSG2	SCN2B	PRDM5		
DSP	SCN3B	PRKG1		
EMD	SCN4B	SKI		
EYA4	SCN5A	SLC2A10		
FHL1	SNTA1	SMAD3		
FKRP	SOS1	SMAD4		
FKTN	TAZ	TGFB2		
FLNC	TBX20	TGFB3		
GATAD1	TBX5	TGFBR1		
GLA	TCAP	TGFBR2		
GPD1L	TECRL	TNXB		
HCN4	TGFB3	ZNF469		
JPH2	TMEM43			
JUP	TNNC1			
KCND3	TNNI3			
KCNE1	TNNT2			
KCNE2	TPM1			
KCNE3	TRDN			
KCNH2	TRPM4			
KCNJ2	TTN			
KCNJ5	TTR			
KCNJ8	TXNRD2			
KCNQ1	VCL			

FCSNext 5 genes
APOA5
APOC2
GPIHBP1
LMF1
LPL

Sitosterolemia 2 genes
ABCG5
ABCG8

Additional genes available for CustomNext-Cardio
ABCA1
APOC3
APOE
CYP27A1
GAA
GATA4
JAG1
LCAT
LIPA
TBX1



CustomNext-Cardio allows you to choose your own combination of up to 167 genes.

* Optional

Moving Science Forward

Family Variant Testing

Testing for all blood relatives is available within 90 days of the original report if the proband was tested at Ambry. Family testing is done via specific site analysis for pathogenic or likely pathogenic variants. (Excludes SNP array and applies to single gene, panel or exome testing.)

Flexible Sample Options

Ambry accepts blood, saliva and other sample types to perform genetic testing.

Sample for Life

We periodically review variants and let you know when there is updated information, such as a reclassification. This is part of our commitment to finding answers.

Additional Genes for Better Care

Post-reporting, clinicians can add-on additional cardiology genes or panels to be analyzed at no cost, within 90 days of the original cardiogenetic test and without the need for a new sample.

About Ambry



Ambry Genetics, a subsidiary of REALM IDx, Inc., excels at translating scientific research into clinically actionable test results based on a deep understanding of the human genome and the biology behind genetic disease. Ambry has an unparalleled track record of discoveries over 20 years and a database that continually expands through collaboration with academic, corporate and pharmaceutical partners. Being first to market with innovative products and comprehensive analysis, Ambry enables clinicians to confidently inform patient health decisions. For more information, please visit ambrygen.com.

References:

- Centers for Disease Control and Prevention. "Heart Disease Prevalence." National Center for Health Statistics. <https://www.cdc.gov/nchs/hsr/topics/heart-disease-prevalence.htm>. August 2022.
- Zeng L, Talukdar HA, Koplev S, et al. Contribution of Gene Regulatory Networks to Heritability of Coronary Artery Disease. *J Am Coll Cardiol*. 2019;73(23):2946-2957. doi:10.1016/j.jacc.2019.03.520.



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