Ambry Genetics®

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®)**

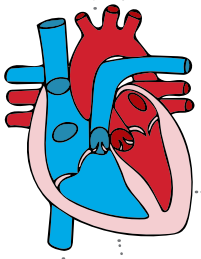
CARDIOMYOPATHIES

Hypertrophic Cardiomyopathy **(HCMNext®)**

Dilated Cardiomyopathy **(DCMNext®)***

Arrhythmogenic Right Ventricular Cardiomyopathy **(ARVCNext)**

Cardiomyopathies **(CMNext®, CardioNext)**



FAMILIAL HYPERCHOLESTEROLEMIA AND LIPID DISORDERS

Familial Hypercholesterolemia **(FHNext®)**

Sitosterolemia **(Sitosterolemia Panel)**

Familial Chylomicronemia Syndrome **(FCSNext)**

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

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)**

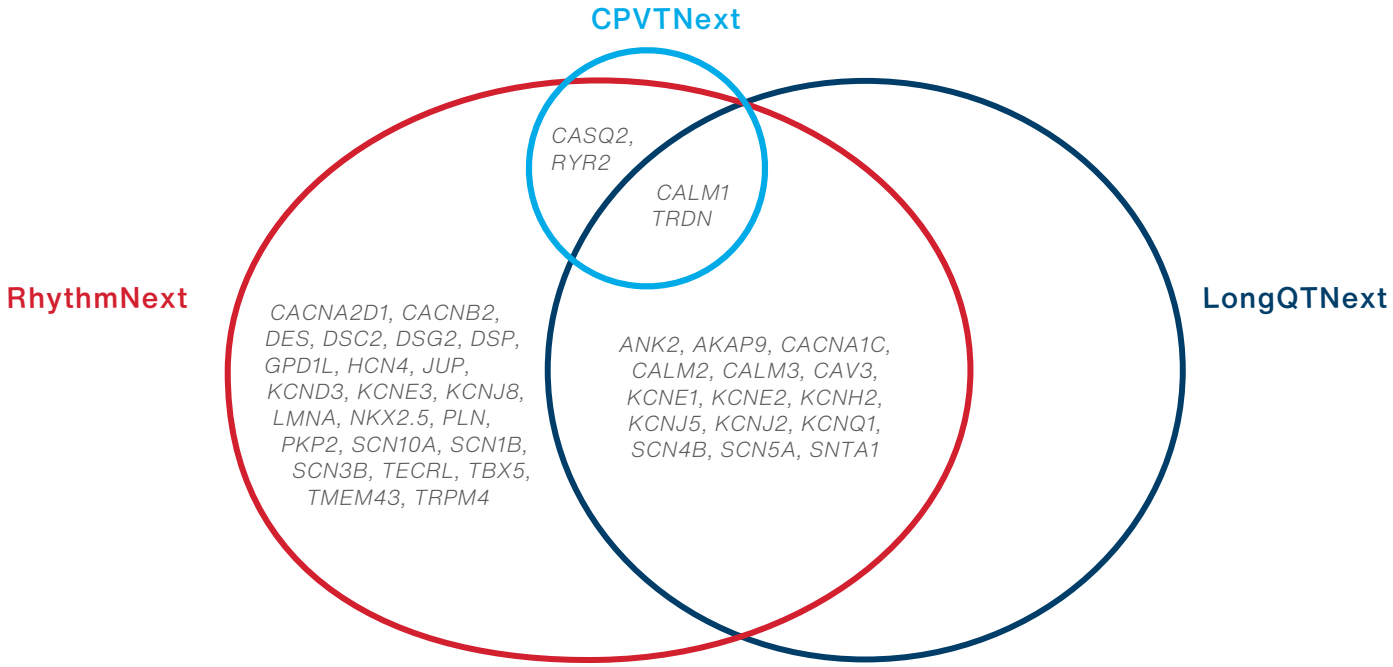
Ehlers-Danlos Syndrome **(TAADNext)**

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

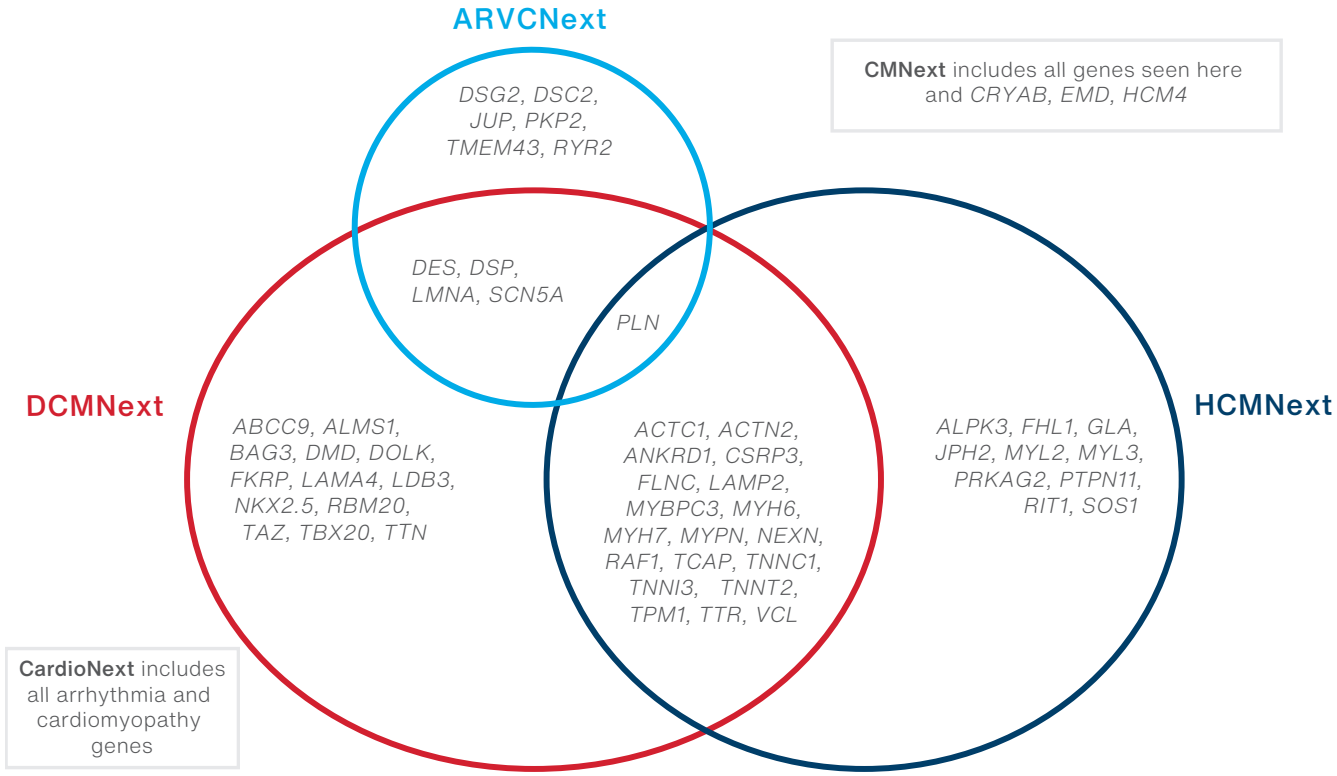
Targeted Panels Gene Comparison

All panels have a turnaround time of 2-3 weeks

ARRHYTHMIAS



CARDIOMYOPATHIES



CMNext includes all genes seen here and *CRYAB, EMD, HCM4*

CardioNext includes all arrhythmia and cardiomyopathy genes

Comprehensive, Targeted, and Custom Panels Gene Lists

All panels have a turnaround time of 2-3 weeks

CardioNext
92 genes

ABCC9	*LAMA4*
ACTC1	*LAMP2*
ACTN2	*LDB3*
AKAP9	*LMNA*
ALMS1	*MYBPC3*
ALPK3	*MYH6*
ANK2	*MYH7*
ANKRD1	*MYL2*
BAG3	*MYL3*
CACNA1C	*MYOZ2*
CACNA2D1	*MYPN*
CACNB2	*NEXN*
CALM1	*NKX2.5*
CALM2	*PKP2*
CALM3	*PLN*
CASQ2	*PRKAG2*
CAV3	*PTPN11*
CRYAB	*RAF1*
CSRP3	*RBM20*
DES	*RIT1*
DMD	*RYR2*
DOLK	*SCN10A*
DSC2	*SCN1B*
DSG2	*SCN2B*
DSP	*SCN3B*
EMD	*SCN4B*
EYA4	*SCN5A*
FHL1	*SNTA1*
FKRP	*SOS1*
FKTN	*TAZ*
FLNC	*TBX20*
GATAD1	*TBX5*
GLA	*TCAP*
GPD1L	*TECRL*
HCN4	*TGFB3*
JPH2	*TMEM43*
JUP	*TNNC1*
KCND3	*TNNI3*
KCNE1	*TNNT2*
KCNE2	*TPM1*
KCNE3	*TRDN*
KCNH2	*TRPM4*
KCNJ2	*TTN*
KCNJ5	*TTR*
KCNJ8	*TXNRD2*
KCNQ1	*VCL*

TAADNext
35 genes

ACTA2
BGN
CBS
CHST14
COL1A1
COL1A2
COL3A1
COL5A1
COL5A2
EFEMP2
FBN1
FBN2
FKBP14
FLNA
FOXE3
LOX
MAT2A
MED12
MFAP5
MYH11
MYLK
NOTCH1
PLOD1
PRDM5
PRKG1
SKI
SLC2A10
SMAD3
SMAD4
TGFB2
TGFB3
TGFBR1
TGFBR2
TNXB
ZNF469

NoonanNext
18 genes

BRAF
CBL
HRAS
KRAS
LZTR1
MAP2K1
MAP2K2
NF1
NRAS
PPP1CB
PTPN11
RAF1
RASA1
RIT1
SHOC2
SOS1
SOS2
SPRED1

FHNext
4 genes + SNP

APOB
LDLR
LDLRAP1
PCSK9
*SLCO1B1 (c.521T>C)**

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

No-cost Family Variant Testing

No-cost testing for all blood relatives is now 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.

Updates to Variant Classification

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 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:

1. Centers for Disease Control and Prevention. “Heart Disease Prevalence.” National Center for Health Statistics. https://www.cdc.gov/nchs/hus/topics/heart-disease-prevalence.htm. August 2022.
2. 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.

Scan this QR code to learn more

One Enterprise, Aliso Viejo, CA 92656 USA Toll Free +1.866.262.7943 Fax +1.949.900.5501 **ambrygen.com**

MKT-SPEC-BRO-20026-EN v6 07.01.24