Ambry Genetics®
A Konica Minolta Company

Cardiogenetics Testing

REFERENCE GUIDE

December 2018

Why Choose Ambry

More than 1 in 200 people have an inherited cardiovascular condition. Ambry's mission is to provide the most advanced genetic testing information available to help you identity 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)

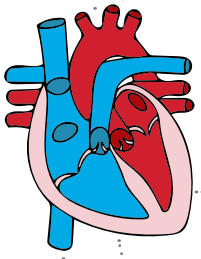
CARDIOMYOPATHIES

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)

Comprehensive Lipid Menu (CustomNext-*Cardio*)

Familial Chylomicronemia Syndrome (FCSNext)

OTHER CARDIO CONDITIONS

Transthyretin Amyloidosis (TTR)

Noonan Syndrome (NoonanNext)

Hereditary Hemorrhagic Telangiectasia (HHTNext)

THORACIC AORTIC ANEURYSMS AND DISSECTIONS

Thoracic Aneurysms and Dissections, aortopathies (TAADNext)

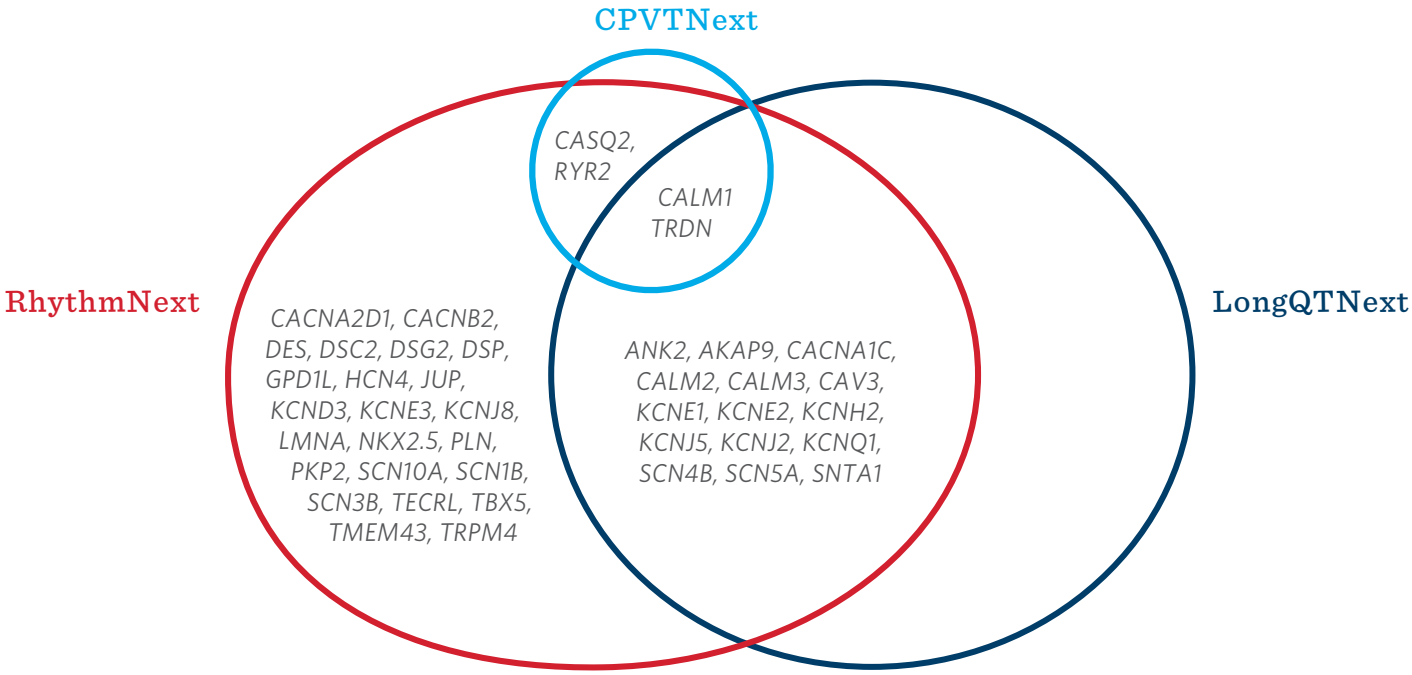
Marfan Syndrome (TAADNext)

Ehlers-Danlos Syndrome (TAADNext)

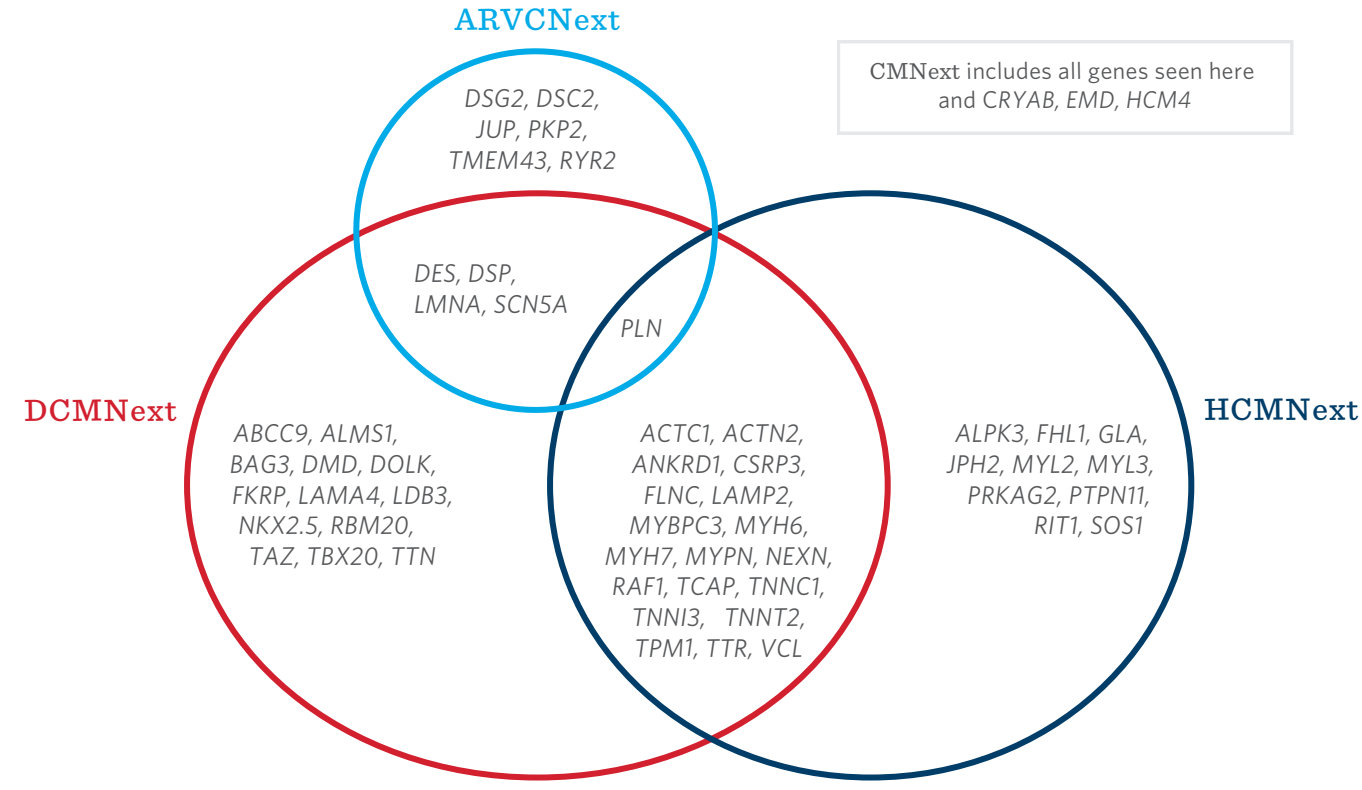
Targeted Panels Gene Comparison

ALL PANELS HAVE A TURNAROUND TIME OF 2-3 WEEKS

ARRHYTHMIAS



CARDIOMYOPATHIES



Comprehensive, Targeted and Lipid Panel 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

CustomNext-*Cardio*
Up to 18 additional lipid genes

ABCA1
ABCG5
ABCG8
APOA1
APOA5
APOB
APOC2
APOC3
APOE
CYP27A1
GPIHBP1
LCAT
LDLR
LDLRAP1
LIPA
LMF1
LPL
PCSK9

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

** Optional*

Over 1 Million Tests Completed

MOVING SCIENCE FORWARD

Free Family Member Testing

Free testing for ALL family members 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.)

Purposeful Confirmatory Testing

Many labs validate their tests based on certain limited studies. That's why we led the largest study of its kind (20,000 cases) guiding us to utilize confirmatory testing when we see specific well-defined thresholds. Our mission is to get it right the first time.

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.

About Ambry

Just as no two fingerprints are alike, the way disease presents itself in every individual is different. Since 1999, our mission has always been about understanding disease better, so treatments and cures can be found faster. Every sample that arrives in our lab is viewed as a person with a life and a story that is unique to only them. By providing advanced confirmation genetic testing for inherited and non-inherited diseases, we can help you make more informed and responsible medical management decisions with your patients.

Visit ambrygen.com for more information.

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