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 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**

- Transthyretin Amyloidosis (TTR)
- Noonan Syndrome (NoonanNext)
- Hereditary Hemorrhagic Telangiectasia (HHTNext)

**FAMILIAL HYPERCHOLESTEROLEMIA AND LIPID DISORDERS**

- Familial Hypercholesterolemia (FHNext)
- Sitosterolemia (Sitosterolemia Panel)
- Comprehensive Lipid Menu (CustomNext-Cardio)
- Familial Chylomicronemia Syndrome (FCSNext)

**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**

**CPVTNext**
- CASQ2, RYR2

**LongQTNext**
- ANK2, AKAP9, CACNA1C, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ5, KCNJ2, KCNQ1, SCN4B, SCNSA, SNTA1

**RhythmNext**
- CACNA2D1, CACNB2, DES, DSC2, DSG2, DSP, GPD1L, HCN4, JUP, KCNND3, KCNE3, KCNJ8, LMNA, NNX2.5, PLN, PKP2, SCN10A, SCN1B, SCN3B, TECRL, TBX5, TMEM43, TRPM4

**CARDIOMYOPATHIES**

**ARVCNNext**
- DSG2, DSC2, JUP, PKP2, TMEM43, RYR2

**CMNext**
- ACTC1, ACTN2, ANKRD1, CSRP3, FLCN, LAMP2, MYBPC3, MYH6, MYH7, MYPN, NEXN, RAF1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL

**DCMNext**
- ABCC9, ALMS1, BAG3, DMD, DOLK, FKRP, LAMA4, LDB3, NNX2.5, RBM20, TAZ, TBX20, TTN

**HCMNext**
- ALPK3, FHL1, GLA, JPH2, MYL2, MYL3, PRKAG2, PTPN11, RIT1, SOS1

CMNext includes all genes seen here and CRYAB, EMD, HCM4
# Comprehensive, Targeted and Lipid Panel Gene Lists

**CardioNext**  
92 genes

- ABCG9
- ACTC1
- ACTN2
- AKAP9
- ALMS1
- ALPK3
- ANK2
- ANKRD1
- BAG3
- CACNA1C
- CACNA2D1
- CACNB2
- CALM1
- CALM2
- CALM3
- CASQ2
- CAV3
- CRYAB
- CSRP3
- DES
- DMD
- DOLK
- DSC2
- DSG2
- DSP
- EMD
- EYA4
- FHL1
- FKBP14
- FLNC
- GLA
- GP1D1
- HCN4
- JPH2
- JUP
- KCN3
- KCNE1
- KCNE2
- KCNE3
- KCNH2
- KCNJ2
- KCNJ5
- KCNJ8
- KCNQ1
- LAMA4
- LAMP2
- LDB3
- LMNA
- MYBPC3
- MYH6
- MYH7
- MYL2
- MYL3
- MYOZ2
- MYPN
- NEXN
- NNX2.S
- PKP2
- PLN
- PRKAG2
- PTPN11
- RAF1
- RBBM20
- RIT1
- RYR2
- SCN10A
- SCN1B
- SCN2B
- SCN3B
- SCN4B
- SCNSA
- SNTA1
- SOS1
- TAZ
- TBX20
- TBX5
- TCAP
- TECRL
- TGFBS
- TMEM43
- TNNC1
- TNNI3
- TNNT2
- TPM1
- TRDN
- TRPM4
- TTN
- TTR
- TXNRD2
- VCL

**TAADNext**  
35 genes

- ACTA2
- BGN
- CBS
- CHST14
- COL1A1
- COL1A2
- COL3A1
- COL5A1
- COL5A2
- EEFMP2
- FBN1
- FBN2
- FKBP14
- FLNA
- FOXE3
- LOX
- MAT2A
- MED12
- MAFAP5
- MYH11
- MYLK
- NOTCH1
- PLOD1
- PRDM5
- PRKG1
- SKI
- SLC2A10
- SMAD3
- SMAD4
- TGFBS2
- TGFBS3
- 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

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*Optional*

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