Overview

Test Description

This panel evaluates 171 genes associated with hereditary cardiovascular conditions, including cardiomyopathies and channelopathies, inherited aortopathies, hereditary lipidemias, thrombophilia, and syndromic and neuromuscular conditions with significant cardiac involvement.

Genetics Information

This test utilizes next-generation sequencing to detect single nucleotide variants, insertions and deletions up to 20 bp, and copy number variants in genes associated with hereditary cardiovascular conditions.

Indications For Testing

A relevant personal and/or family history suggestive of a hereditary cardiovascular condition, including cardiomyopathy, arrhythmia, unexplained syncope or sudden cardiac death, thoracic aortic aneurysm or dissection, severe or early-onset dyslipidemia, or venous thromboembolism.

Clinical Descriptions

Hereditary cardiovascular conditions span a broad range of genetic disorders affecting the structure and function of the heart and vasculature. Cardiomyopathies — including hypertrophic (HCM), dilated (DCM), arrhythmogenic (ARVC), and left ventricular non-compaction (LVNC) — are structural and functional disorders of the myocardium that may cause heart failure, arrhythmia, and sudden cardiac death; many present with normal or near-normal findings early in the disease course. Channelopathies such as long QT syndrome, Brugada syndrome, short QT syndrome, and catecholaminergic polymorphic ventricular tachycardia (CPVT) are electrophysiologic disorders that predispose to potentially fatal arrhythmias in the absence of structural abnormality.

Inherited aortopathies on this panel include isolated thoracic aortic aneurysm and dissection (TAAD) as well as syndromic aortopathies such as Marfan syndrome and Loeys-Dietz syndrome. Genes associated with connective tissue disorders that confer vascular fragility, including vascular Ehlers-Danlos syndrome, are also included given the risk of arterial aneurysm and dissection. Hereditary lipidemias include familial hypercholesterolemia, primary hypertriglyceridemias and chylomicronemia syndromes, and disorders of HDL metabolism, carrying risk of premature atherosclerotic cardiovascular disease and, in severe cases, pancreatitis. The panel also includes genes associated with factor V Leiden-related and prothrombin-related thrombophilia.

A subset of genes is associated with syndromic and multisystem conditions in which cardiac involvement is a

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Clinical Descriptions

primary or defining feature. These include the RASopathies (Noonan syndrome, cardiofaciocutaneous syndrome, and related conditions); metabolic and storage disorders such as Fabry disease, Danon disease, Pompe disease, and hereditary transthyretin amyloidosis; fatty acid oxidation and organic acid disorders; mitochondrial cardiomyopathies; and muscular dystrophies and myopathies with cardiac manifestations, including Duchenne/Becker and Emery-Dreifuss muscular dystrophy. Inheritance across the panel may be autosomal dominant, autosomal recessive, X-linked, or mitochondrial.

Identification of a pathogenic variant can establish a molecular diagnosis, guide condition-specific management and surveillance, and facilitate cascade testing in at-risk family members.

Test Details

Genes Tested

ABCA1, ABCC9, ABCG5, ABCG8, ACAD9, ACADVL, ACTA2, ACTC1, ACTN2, ADAMTS10, AGL, ALMS1, ALPK3, ANGPTL3, ANK2, APOA1, APOA5, APOB, APOC2, APOE, BAG3, BAG5, BGN, BMP10, BRAF, CACNA1C, CACNA1D, CALM1, CALM2, CALM3, CASQ2, CAV3, CBS, CDH2, CETP, COL3A1, COL5A1, COL5A2, CPT2, CREB3L3, CRYAB, CSRP3, CYP27A1, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, EFEMP2, ELAC2, EMD, F2, F5, FBN1, FBN2, FHL1, FKRP, FKTN, FLII, FLNA, FLNC, FOXE3, GAA, GLA, GPD1, GPIHBP1, HCN4, HRAS, JPH2, JUP, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, KRAS, LAMP2, LCAT, LDB3, LDLR, LDLRAP1, LIPA, LIPG, LMF1, LMNA, LMOD2, LOX, LPL, LRP6, LZTR1, MAP2K1, MAP2K2, MED12, MFAP5, MRAS, MTO1, MTTP, MYBPC3, MYH11, MYH7, MYL2, MYL3, MYL4, MYLK, MYLK3, MYPN, MYZAP, NEXN, NKX2-5, NOTCH1, NRAP, NRAS, PCCA, PCCB, PCSK9, PKP2, PLEKHM2, PLN, PLOD1, PNPLA2, PPA2, PPARG, PPCS, PPP1R13L, PRDM16, PRKAG2, PRKG1, PTPN11, RAF1, RBM20, RIT1, RPL3L, RYR2, SAR1B, SCN5A, SGCD, SHOC2, SKI, SLC22A5, SLC2A10, SLC4A3, SMAD2, SMAD3, SMAD4, SOS1, SOS2, SYNE2, TAFAZZIN, TBX20, TBX5, TCAP, TECRL, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TTN, TTR, VCL

Methods & Limitations

Extracted DNA is enriched for targeted regions and then sequenced using the Helix Exome+ (R) assay on an Illumina DNA sequencing system. Data is then aligned to a modified version of GRCh38 and all genes are analyzed using the MANE transcript and MANE Plus Clinical transcript, when available. Small variant calling is completed using a customized version of Sentieon's DNAseq software, augmented by a proprietary small variant caller for difficult variants. Copy number variants (CNVs) are then called using a proprietary bioinformatics pipeline based on depth analysis with a comparison to similarly sequenced samples. Interpretation is based upon guidelines published by the American College of Medical Genetics and Genomics (ACMG) and the Association for Molecular Pathology (AMP) or their modification by ClinGen Variant Curation Expert Panels when available. Interpretation is limited to the transcripts indicated on the report, +/- 10 bp into intronic regions, except as noted below. Helix variant classifications include pathogenic, likely

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pathogenic, variant of uncertain significance (VUS), likely benign, and benign. Variants classified as pathogenic, likely pathogenic, or VUS are included in the report. All reported variants (except for VUSs with limited evidence of pathogenicity) are confirmed through secondary manual inspection of DNA sequence data or orthogonal testing. Benign and likely benign variants are not reported but are available upon request. Gene-disease associations are routinely reviewed and may change over time. Risk estimations and management guidelines included in this report are based on analysis of primary literature and recommendations of applicable professional societies, and should be regarded as approximations.

Based on validation studies, this assay delivers > 99% sensitivity and specificity for single nucleotide variants and insertions and deletions (indels) up to 20 bp. Larger indels and complex variants are also reported but sensitivity may be reduced. Based on validation studies, this assay delivers > 99% sensitivity to multi-exon CNVs and > 90% sensitivity to single-exon CNVs. This test may not detect variants in challenging regions (such as short tandem repeats, homopolymer runs, and segment duplications), sub-exonic CNVs, chromosomal aneuploidy, or variants in the presence of mosaicism. Phasing will be attempted and reported, when possible. Structural rearrangements such as inversions, translocations, complex rearrangements, and gene conversions are not tested in this assay unless explicitly indicated. Additionally, deep intronic, promoter, and enhancer regions may not be covered. It is important to note that this assay cannot detect all variants known to increase disease risk, and that a negative result does not guarantee that the tested individual does not carry a rare, undetectable variant in genes analyzed. Any potential incidental findings outside of these genes and conditions will not be identified, nor reported. The results of a genetic test may be influenced by various factors, including bone marrow transplantation, blood transfusions, or in rare cases, hematolymphoid neoplasms.

Gene Specific Notes:
ABCA1: Analysis for ABCA1 exon 40 will not be performed. *ALPK3*: Sensitivity in ALPK3 exon 1 may be reduced. *BRAF*: Sensitivity in BRAF exon 1 may be reduced. *CDH2*: Sensitivity in CDH2 exon 1 may be reduced. *F2*: Results limited to F2:c.*97G>A. *F5*: Results limited to F5:c.1601G>A (p.Arg534Gln). FOXE3: Analysis begins at chr1:47416567 (GRCh38) and excludes the first quarter of exon 1. *KCNQ1*: Sensitivity in KCNQ1 exon 1 may be reduced. *LDB3*: Sensitivity in LDB3 (NM_007078.3) exons 4, 8, and 9 may be reduced. LDLR: Includes CNV detection in the LDLR promoter. *MAP2K2*: Sensitivity in MAP2K2 exon 1 may be reduced. *MYZAP*: Sensitivity in MYZAP exons 1 and 4 may be reduced. *NRAP*: Sensitivity in NRAP exons 6, 10, 13, 16, 23, 25, 26, 34, 36, and 37 may be reduced. *PLEKHM2*: Sensitivity in PLEKHM2 exons 1, 8, and 16 may be reduced. *PRDM16*: Analysis for PRDM16 exon 1 will not be performed. PRKAG2: Sensitivity in PRKAG2 exon 5 may be reduced. *RPL3L*: Sensitivity in RPL3L exons 2 and 3 may be reduced. *SAR1B*: Sensitivity in SAR1B exon 4 may be reduced. SOS2: Sensitivity in SOS2 exon 1 may be reduced. *TGFBR1*: Analysis for TGFBR1 exon 1 will not be performed. *TTN*: Analysis for TTN exons 172 to 197 will not be performed.

Disclaimer

This test was developed and validated by Helix, Inc. This test has not been cleared or approved by the United States Food and Drug Administration (FDA). The Helix laboratory is accredited by the College of American

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Disclaimer

Pathologists (CAP) and certified under the Clinical Laboratory Improvement Amendments (CLIA #: 05D2117342) to perform high-complexity clinical tests. This test is used for clinical purposes. It should not be regarded as investigational use only or for research use only.

Interpretation

All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Reclassification

Helix reviews variant classifications annually when they arise in routine processes and upon request from providers. The timing of re-review depends on clinical risk. Providers can request a variant re-review by contacting Helix Customer Support. If a classification by Helix is updated, Helix identifies affected past patients and issues revised reports. Updated results are communicated to providers prior to results being uploaded to the EHR, and patients are notified through the EHR patient portal.

Variant Evaluation

Variant classification is performed using the guidelines set forth by the American College of Medical Genetics and Genomics and the Association for Molecular Pathology, with modifications as suggested by domain specific Expert Panels of the Clinical Genome Resource (ClinGen) when available. Variant pathogenicity is categorized as benign, likely benign, variant of uncertain significance (VUS), likely pathogenic, or pathogenic.

Laboratory Details

Turnaround Time - Standard	Typically 6 to 21 days	**Turnaround Time - Requery (SOQO®)**	Typically ≤ 5 days
Available in NY State	Yes	**Procedure Code**	H01426-2 (Helix)
Test Classification	This test was developed, and its performance characteristics determined, by Helix, Inc. in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.		
Regulatory Information	• CLIA Complexity: High • Test Classification: Non-Waived/ Laboratory Developed Test		
CLIA Category	Chemistry / Routine Chemistry		

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Laboratory Details

Performing Laboratory Information	• CLIA Laboratory Number: 05D2117342 • Laboratory Hours of Operation: Monday-Saturday (7AM-10:30PM PST) • Address: 10170 Sorrento Valley Road, Suite 100, San Diego, CA 92121 • Helix Customer Support: (844) 211-2070 • Email: support@helix.com

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Conditions List

- 3-methylglutaconic aciduria type 5
- Abetalipoproteinemia
- Agenesis of corpus callosum, cardiac, ocular, and genital syndrome (ACOGS)
- Alstrom syndrome
- Andersen-Tawil syndrome
- Aneurysms-osteoarthritis syndrome (Loeys-Dietz syndrome type 3)
- Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities
- Arrhythmogenic right ventricular cardiomyopathy
- Arrhythmogenic right ventricular cardiomyopathy with woolly hair
- Arterial tortuosity syndrome
- Atrial fibrillation
- Barth syndrome
- Brugada syndrome
- Cantu syndrome
- Cardiofaciocutaneous syndrome
- Carnitine palmitoyltransferase II deficiency
- Catecholaminergic polymorphic ventricular tachycardia
- Caveolinopathy
- Cerebrotendinous xanthomatosis
- Chylomicron retention disease (CMRD) / Anderson disease
- Classic homocystinuria
- Combined lipase deficiency
- Combined oxidative phosphorylation defect type 17
- Congenital contractural arachnodactyly
- Congenital myopathy
- Costello syndrome
- Cutis laxa, type 1B
- Danon disease
- Dilated cardiomyopathy
- Distal myopathy
- DK1-congenital disorder of glycosylation
- Ehlers-Danlos syndrome, classic type
- Ehlers-Danlos syndrome, kyphoscoliotic type 1
- Ehlers-Danlos syndrome, vascular type
- Emery-Dreifuss muscular dystrophy
- Fabry disease
- Factor V deficiency
- Factor V Leiden Thrombophilia / Thrombophilia due to activated protein C resistance
- Familial apolipoprotein C-II deficiency
- Familial hypercholesterolemia
- Familial hypertriglyceridemia
- Familial hypertrophic cardiomyopathy, type 27
- Familial hypobetalipoproteinemia
- Familial lipoprotein lipase deficiency
- Familial thoracic aortic aneurysm and aortic dissection
- Fatal infantile hypertonic myofibrillar myopathy
- Glycogen storage disease type III
- Glycogen storage-related Wolff-Parkinson-White with or without hypertrophic cardiomyopathy
- Hereditary transthyretin-related amyloidosis
- Holt-Oram syndrome
- Hutchinson-Gilford progeria syndrome
- Hyperalphalipoproteinemia
- Hyperlipoproteinemia, type 1D
- Hyperlipoproteinemia, type 5
- Hyperlipoproteinemia, type III
- Hypertrophic cardiomyopathy
- Hypoplastic left heart
- Intrinsic cardiomyopathy
- Jervell and Lange-Nielsen syndrome
- Juvenile polyposis / Hereditary hemorrhagic telangiectasia syndrome
- LCAT deficiency / Fish-eye disease / Norum disease
- Left ventricular noncompaction

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- Limb-girdle muscular dystrophy
- Lipodystrophy
- Loeys-Dietz syndrome, type 1
- Loeys-Dietz syndrome, type 2
- Loeys-Dietz syndrome, type 4
- Loeys-Dietz syndrome, type 6
- Long QT syndrome
- Lysosomal acid lipase (LAL) deficiency
- Marfan syndrome
- Meester-Loeys syndrome
- Melnick-Needles syndrome
- Mitochondrial complex I deficiency
- Mitochondrial complex V deficiency nuclear type 2
- Mitochondrial hypertrophic cardiomyopathy with lactic acidosis
- Multisystemic smooth muscle dysfunction syndrome
- Muscular dystrophy
- Muscular dystrophy-dystroglycanopathy
- Myhre syndrome
- Myofibrillar myopathy
- Myopathy
- Naxos disease
- Neutral lipid storage myopathy
- Noonan syndrome
- NOTCH1-related Adams-Oliver syndrome (AOS) spectrum disorder
- Otopalatodigital syndrome type 2
- Periventricular nodular heterotopia
- Polymicrogyria with or without vascular-type Ehlers-Danlos syndrome
- Pompe disease
- Primary carnitine deficiency disease
- Primary hypoalphalipoproteinemia
- Progeroid and marfanoid aspect-lipodystrophy syndrome
- Progressive muscular dystrophy
- Propionic acidemia
- Prothrombin deficiency
- Prothrombin thrombophilia / Prothrombin-related thrombophilia
- Restrictive cardiomyopathy
- Rienhoff syndrome (Loeys-Dietz syndrome type 5)
- Short QT syndrome
- Shprintzen-Goldberg syndrome
- Sinoatrial node dysfunction and deafness
- Sinus node dysfunction or bradycardia
- Sitosterolemia
- Skeletal myopathy
- Sudden cardiac failure
- Tangier disease
- Tibial muscular dystrophy
- Timothy syndrome
- Transient infantile hypertriglyceridemia
- Very long chain acyl-CoA dehydrogenase deficiency
- Weill-Marchesani syndrome 1
- Weill-Marchesani syndrome 2
- Wolff-Parkinson-White syndrome

In addition, one or more of the genes on this panel are associated with other conditions for which a predisposition to, or carrier status of, may be identified:

- Acromicric dysplasia
- Aldosterone-producing adenoma with seizures and neurological abnormalities
- Alzheimer's disease*
- Anterior segment dysgenesis
- Atrial septal defects
- Cardiac valvular dysplasia
- Cataracts
- Charcot-Marie-Tooth disease
- Complex neurodevelopmental disorder

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- Congenital heart disease
- Congenital primary aphakia
- Conotruncal heart malformations
- Hereditary gingival fibromatosis
- Intestinal pseudoobstruction, neuronal
- LZTR1-related schwannomatosis
- MED12-related intellectual disability syndrome
- Megacystis-microcolon-intestinal hypoperistalsis syndrome 2
- Metachondromatosis
- Multiple self-healing squamous epithelioma
- Spondyloepimetaphyseal dysplasia
- Stiff skin syndrome
- Structural heart defects
- Terminal osseous dysplasia-pigmentary defects syndrome

** Helix does not report on risk for Alzheimer's disease as part of this test.*

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Targeted Genes

The following applies to the Helix Comprehensive Cardiovascular Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequences on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants, as listed below. Next-generation sequencing is performed to test for the presence of small variants and copy number variants in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from October 2026 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. For questions regarding genes, reference transcripts, or specific regions covered, contact Helix Customer Support at (844) 211-2070.

Genomic Build: GRCh38

Gene	Transcript	Special Gene Notes
ABCA1	NM_005502.4	Analysis for ABCA1 exon 40 will not be performed
ABCC9	NM_020297.4	–
ABCG5	NM_022436.3	–
ABCG8	NM_022437.3	–
ACAD9	NM_014049.5	–
ACADVL	NM_000018.4	–
ACTA2	NM_001613.4	–
ACTC1	NM_005159.5	–
ACTN2	NM_001103.4	–
ADAMTS10	NM_030957.4	–
AGL	NM_000642.3	–
ALMS1	NM_001378454.1	–
ALPK3	NM_020778.5	Sensitivity in ALPK3 exon 1 may be reduced
ANGPTL3	NM_014495.4	–
ANK2	NM_001148.6	–
APOA1	NM_000039.3	–
APOA5	NM_001371904.1	–

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Gene	Transcript	Special Gene Notes
APOB	NM_000384.3	–
APOC2	NM_000483.5	–
APOE	NM_000041.4	–
BAG3	NM_004281.4	–
BAG5	NM_001015048.3	–
BGN	NM_001711.6	–
BMP10	NM_014482.3	–
BRAF	NM_004333.6; NM_001374258.1	Sensitivity in BRAF exon 1 may be reduced
CACNA1C	NM_000719.7; NM_001167623.2	–
CACNA1D	NM_001128840.3; NM_000720.4	–
CALM1	NM_006888.6	–
CALM2	NM_001743.6	–
CALM3	NM_005184.4	–
CASQ2	NM_001232.4	–
CAV3	NM_033337.3	–
CBS	NM_000071.3	–
CDH2	NM_001792.5	Sensitivity in CDH2 exon 1 may be reduced
CETP	NM_000078.3	–
COL3A1	NM_000090.4	–
COL5A1	NM_000093.5	–
COL5A2	NM_000393.5	–
CPT2	NM_000098.3	–
CREB3L3	NM_032607.3	–
CRYAB	NM_001289808.2	–
CSRP3	NM_003476.5	–
CYP27A1	NM_000784.4	–
DES	NM_001927.4	–
DMD	NM_004006.3	–
DNAJC19	NM_145261.4	–

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Gene	Transcript	Special Gene Notes
DOLK	NM_014908.4	–
DSC2	NM_024422.6	–
DSG2	NM_001943.5	–
DSP	NM_004415.4	–
DTNA	NM_001386795.1	–
EFEMP2	NM_016938.5	–
ELAC2	NM_018127.7	–
EMD	NM_000117.3	–
F2	NM_000506.5	Results limited to F2:c.*97G>A
F5	NM_000130.5	Results limited to F5:c.1601G>A (p.Arg534Gln)
FBN1	NM_000138.5	–
FBN2	NM_001999.4	–
FHL1	NM_001159699.2; NM_001159702.3	–
FKRP	NM_024301.5	–
FKTN	NM_001079802.2	–
FLII	NM_002018.4	–
FLNA	NM_001110556.2	–
FLNC	NM_001458.5	–
FOXE3	NM_012186.3	Analysis begins at chr1:47416567 (GRCh38) and excludes the first quarter of FOXE3 exon 1
GAA	NM_000152.5	–
GLA	NM_000169.3	–
GPD1	NM_005276.4	–
GPIHBP1	NM_178172.6	–
HCN4	NM_005477.3	–
HRAS	NM_005343.4; NM_176795.5	–
JPH2	NM_020433.5	–
JUP	NM_002230.4	–

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Gene	Transcript	Special Gene Notes
KCNE1	NM_000219.6	–
KCNE2	NM_172201.2	–
KCNH2	NM_000238.4	–
KCNJ2	NM_000891.3	–
KCNQ1	NM_000218.3	Sensitivity in KCNQ1 exon 1 may be reduced
KRAS	NM_004985.5; NM_033360.4	–
LAMP2	NM_002294.3	–
LCAT	NM_000229.2	–
LDB3	NM_007078.3; NM_001368067.1	Sensitivity in LDB3 (NM_007078.3) exons 4, 8, and 9 may be reduced
LDLR	NM_000527.5	Includes CNV detection in the LDLR promoter
LDLRAP1	NM_015627.3	–
LIPA	NM_000235.4	–
LIPG	NM_006033.4	–
LMF1	NM_022773.4	–
LMNA	NM_170707.4; NM_005572.4	–
LMOD2	NM_207163.3	–
LOX	NM_002317.7	–
LPL	NM_000237.3	–
LRP6	NM_002336.3	–
LZTR1	NM_006767.4	–
MAP2K1	NM_002755.4	–
MAP2K2	NM_030662.4	Sensitivity in MAP2K2 exon 1 may be reduced
MED12	NM_005120.3	–
MFAP5	NM_003480.4	–
MRAS	NM_001085049.3	–
MTO1	NM_012123.4	–
MTTP	NM_001386140.1	–
MYBPC3	NM_000256.3	–

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Gene	Transcript	Special Gene Notes
MYH11	NM_002474.3; NM_001040113.2	–
MYH7	NM_000257.4	–
MYL2	NM_000432.4	–
MYL3	NM_000258.3	–
MYL4	NM_002476.2	–
MYLK	NM_053025.4	–
MYLK3	NM_182493.3	–
MYPN	NM_032578.4	–
MYZAP	NM_001018100.5	Sensitivity in MYZAP exons 1 and 4 may be reduced
NEXN	NM_144573.4	–
NKX2-5	NM_004387.4	–
NOTCH1	NM_017617.5	–
NRAP	NM_198060.4	Sensitivity in NRAP exons 6, 10, 13, 16, 23, 25, 26, 34, 36, and 37 may be reduced
NRAS	NM_002524.5	–
PCCA	NM_000282.4	–
PCCB	NM_000532.5	–
PCSK9	NM_174936.4	–
PKP2	NM_001005242.3	–
PLEKHM2	NM_015164.4	Sensitivity in PLEKHM2 exons 1, 8, and 16 may be reduced
PLN	NM_002667.5	–
PLOD1	NM_000302.4	–
PNPLA2	NM_020376.4	–
PPA2	NM_176869.3	–
PPARG	NM_138711.6	–
PPCS	NM_024664.4	–
PPP1R13L	NM_006663.4	–
PRDM16	NM_022114.4	Analysis for PRDM16 exon 1 will not be performed

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Gene	Transcript	Special Gene Notes
PRKAG2	NM_016203.4	Sensitivity in PRKAG2 exon 5 may be reduced
PRKG1	NM_006258.4	–
PTPN11	NM_002834.5	–
RAF1	NM_002880.4	–
RBM20	NM_001134363.3	–
RIT1	NM_006912.6	–
RPL3L	NM_005061.3	Sensitivity in RPL3L exons 2 and 3 may be reduced
RYR2	NM_001035.3	–
SAR1B	NM_016103.4	Sensitivity in SAR1B exon 4 may be reduced
SCN5A	NM_000335.5; NM_001099404.2	–
SGCD	NM_000337.6	–
SHOC2	NM_007373.4	–
SKI	NM_003036.4	–
SLC22A5	NM_003060.4	–
SLC2A10	NM_030777.4	–
SLC4A3	NM_005070.4	–
SMAD2	NM_005901.6	–
SMAD3	NM_005902.4	–
SMAD4	NM_005359.6	–
SOS1	NM_005633.4	–
SOS2	NM_006939.4	Sensitivity in SOS2 exon 1 may be reduced
SYNE2	NM_182914.3	–
TAFAZZIN	NM_000116.5	–
TBX5	NM_181486.4	–
TBX20	NM_001077653.2	–
TCAP	NM_003673.4	–
TECRL	NM_001010874.5	–
TGFB2	NM_003238.6	–
TGFB3	NM_003239.5	–

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Gene	Transcript	Special Gene Notes
TGFBR1	NM_004612.4	Analysis for TGFBR1 exon 1 will not be performed
TGFBR2	NM_003242.6	–
TMEM43	NM_024334.3	–
TMEM70	NM_017866.6	–
TNNC1	NM_003280.3	–
TNNI3	NM_000363.5	–
TNNI3K	NM_015978.3	–
TNNT2	NM_001276345.2	–
TPM1	NM_001018005.2	–
TRDN	NM_006073.4	–
TRIM63	NM_032588.4	–
TTN	NM_001267550.2	Analysis for TTN exons 172 to 197 will not be performed
TTR	NM_000371.4	–
VCL	NM_014000.3	–

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