Cardiovascular Diseases Genetic Testing Program

Description:

We offer comprehensive gene panels designed to diagnose the most common genetic causes of hereditary cardiovascular diseases. Testing is available for congenital heart malformation, cardiomyopathy, arrhythmia, thoracic aortic aneurysm, pulmonary arterial hypertension, Marfan syndrome, and RASopathy/Noonan spectrum disorders. Hereditary cardiovascular disease is caused by variants in many different genes, and may be inherited in an autosomal dominant, autosomal recessive, or X-linked manner. Other than condition-specific panels, we also offer single gene sequencing for any gene on the panels, targeted variant analysis, and targeted deletion/duplication analysis.

Tests Offered:

Arrhythmia Panels

- Comprehensive Arrhythmia Panel (81 genes)
- Atrial Fibrillation (A Fib) Panel (28 genes)
- Atrioventricular Block (AV Block) Panel (7 genes)
- Brugada Syndrome Panel (21 genes)
- Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel (11 genes)
- Long QT Syndrome Panel (19 genes)
- Short QT Syndrome Panel (6 genes)

Connective Tissue Disorders Panel

- Thoracic Aortic Aneurysm Panel (49 genes)
- Marfan Syndrome and MFS Related Disorders Panel (3 genes)

Cardiomyopathy Panels

- Comprehensive Cardiomyopathy Panel (135 genes)
- Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel (18 genes)
- Dilated Cardiomyopathy (DCM) Panel (73 genes)
- Hypertrophic Cardiomyopathy (HCM) Panel (98 genes)
- Left Ventricular Noncompaction (LVNC) Panel (36 genes)
- Restrictive Cardiomyopathy (RCM) Panel (9 genes)

Congenital Heart Disease Panels

- Congenital Heart Disease Panel (186 genes)
- Heterotaxy Panel (113 genes)
- RASopathy/Noonan Spectrum Disorders Panel (31 genes)

Other Panels

- Pulmonary Arterial Hypertension (PAH) Panel (20 genes)

Indications:

Panels:

- Confirmation of genetic diagnosis in a patient with a clinical diagnosis of cardiovascular disease
- Carrier or pre-symptomatic diagnosis identification in individuals with a family history of cardiovascular disease of unknown genetic basis

Gene Specific Sequencing:

- Confirmation of genetic diagnosis in a patient with cardiovascular disease and in whom a specific gene associated diagnosis is suspected

Variant Specific Analysis:

- Pre-symptomatic testing of at-risk siblings and parents for medical management
- Carrier identification in individuals in whom specific variant(s) have been identified in the proband
- Prenatal diagnosis of an at-risk fetus, after confirmation of variant(s) in the parent(s) and by prior arrangement only

Deletion/Duplication Analysis:

- Completion of the diagnostic evaluation in a patient with a clinical diagnosis of cardiovascular disease who has had a negative panel or who is heterozygous for a variant in a gene associated with an autosomal recessive condition.

Cincinnati Children's®
changing the outcome together

Genetics and Genomics Diagnostic Laboratory
CLIA#: 36D0656333
Phone: (513) 636-4474
Fax: (513) 636-4373
Email: LabGeneticCounselors@cchmc.org
www.cincinnatichildrens.org/genetics

Specimen:

Provide at least 3 mL of whole blood in a lavender top (EDTA) tube. Alternatively, 10 mcg of high quality DNA extracted from whole blood in a CLIA lab may be submitted.

For prenatal samples, at least 20 mL of amniotic fluid or at least 30 mg chorionic villi or 2 flasks of cultured cells is needed. 3 mL of maternal blood in lavender top (EDTA) tube is needed for maternal cell contamination studies.

Testing Methodology:

Panels: Our panels are performed on genomic DNA using a PCR-free genome sequencing preparation and sequenced on an Illumina sequencing system with paired-end reads to an average autosomal sequencing depth of at least 30X. Sequence reads are aligned to the human reference genome (build UCSC hg38) and variants are identified and evaluated by a validated in-house developed bioinformatics analysis pipeline that includes the usage of Dragen Germline pipeline and Fabric Genomic Analysis platform.

Gene Specific Sequencing: PCR-based or NGS-based sequencing of entire coding region, intron/exon boundaries of the specified gene

Variant Specific Analysis: Sanger sequencing following PCR amplification of the targeted variant(s) of the specified gene

Deletion/Duplication Analysis: Copy number variant analysis of the gene by comparative genomic hybridization

Results:

Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for the clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

Test Sensitivity:

Analytical Sensitivity: The sensitivity of DNA sequencing is over 99% for the detection of nucleotide base changes, small deletions and insertions in the regions analyzed.

Sequencing Test Limitations: Pathogenic variants may be present in a portion of the genes not covered by this test or in regions with suboptimal data due to homologous issue, polynucleotides, or nucleotide repeats, and therefore may not be identified. Thus, the absence of identified pathogenic variants does not exclude the possibility of a genetic etiology for the patient's symptoms. Certain types of mutations are not detected. Only single base pair changes or small insertions or deletions of DNA are detected. Large deletions, duplications, or rearrangements, mitochondrial genome mutations, repeat expansions, low level mosaicism and many epigenetic defects may not be detected by this test.

Regions of Homology

These gene regions with homology may generate suboptimal data with potential false negative results.

GENE	TRANSCRIPT	CHROM	EXON	EXON_START	EXON_END
ABCC9	NM_020297	chr12	Exon40	21797389	21801181
KCNE1	NM_000219	chr21	Exon3	34458654	34458764
LEFTY2	NM_003240	chr1	Exon3	225939361	225939600
MYH6	NM_002471	chr14	Exon26	23390057	23390446
MYH7	NM_000257	chr14	Exon27	23419845	23420234
NOTCH2	NM_024408	chr1	Exon2	120029906	120029987
ODAD2	NM_018076	chr10	Exon10	27961568	27961715
ODAD2	NM_018076	chr10	Exon9	27968923	27969018
SDHA	NM_004168	chr5	Exon14	254393	254506
TNXB	NM_001365276	chr6	Exon44	32041153	32041450
TNXB	NM_001365276	chr6	Exon43	32041771	32041934
TNXB	NM_001365276	chr6	Exon42	32042012	32042173
TNXB	NM_001365276	chr6	Exon41	32042266	32042362
TNXB	NM_001365276	chr6	Exon39	32042699	32042831
TNXB	NM_001365276	chr6	Exon38	32042951	32043081
TNXB	NM_001365276	chr6	Exon37	32043175	32043318
TNXB	NM_001365276	chr6	Exon36	32043437	32043556
TNXB	NM_001365276	chr6	Exon34	32044007	32044129
TNXB	NM_001365276	chr6	Exon33	32044381	32044716
TTN	NM_001267550	chr2	Exon197	178653427	178653507
TTN	NM_001267550	chr2	Exon196	178653621	178653701
TTN	NM_001267550	chr2	Exon195	178653817	178653897
TTN	NM_001267550	chr2	Exon194	178654012	178654095
TTN	NM_001267550	chr2	Exon192	178654445	178654534
TTN	NM_001267550	chr2	Exon191	178654745	178654828

GENE	TRANSCRIPT	CHROM	EXON	EXON_START	EXON_END
TTN	NM_001267550	chr2	Exon190	178654912	178654995
TTN	NM_001267550	chr2	Exon188	178657687	178657767
TTN	NM_001267550	chr2	Exon187	178657881	178657961
TTN	NM_001267550	chr2	Exon185	178658272	178658355
TTN	NM_001267550	chr2	Exon184	178658468	178658551
TTN	NM_001267550	chr2	Exon183	178658705	178658794
TTN	NM_001267550	chr2	Exon182	178659005	178659088
TTN	NM_001267550	chr2	Exon181	178659172	178659255
TTN	NM_001267550	chr2	Exon179	178661948	178662028
TTN	NM_001267550	chr2	Exon178	178662142	178662222
TTN	NM_001267550	chr2	Exon176	178662533	178662616
TTN	NM_001267550	chr2	Exon175	178662729	178662812
TTN	NM_001267550	chr2	Exon174	178662966	178663055
TTN	NM_001267550	chr2	Exon172	178663433	178663516

Low coverage (<10X) regions

GENE	TRANSCRIPT	CHROM	EXON	EXON_START	EXON_END
PKD2	NM_000297.4	chr4	Exon1	88007845	88007879

Please note: These regions represent the low coverage (<10X) regions identified during our test validation. For specific patient cases, these regions may vary slightly.

Turn-Around Time:

Gene panels: up to 6 weeks
Individual/custom gene sequencing: up to 6 weeks
Targeted analysis: up to 2 weeks
Deletion/Duplication analysis: up to 4 weeks

CPT Codes:

- *APOB* (R3500Q/R3500W) Sequencing Analysis: **81401**
- Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel, Dilated Cardiomyopathy (DCM) Panel, Hypertrophic Cardiomyopathy (HCM) Panel, Left Ventricular Noncompaction (LVNC) Panel, Restrictive Cardiomyopathy (RCM) Panel: **81439**
- Atrial Fibrillation (A Fib) Panel, Brugada Syndrome Panel, Congenital Heart Disease Panel (CHD), Heterotaxy Panel, Long QT Syndrome Panel, Pulmonary Arterial Hypertension (PAH) Panel: 81443Atrioventricular Block (AV Block) Panel: **81404, 81405, 81406**
- Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel: **81403, 81405, 81408**
- Comprehensive Arrhythmia Panel: **81443**
- Comprehensive Cardiomyopathy Panel: **81439**
- *FBN1* Full Mutation Analysis: **81408**
- *LDLR* Gene Sequencing: **81406**
- Marfan Syndrome and MFS Related Disorders Panel: **81405 x2, 81408**
- RASopathy/Noonan Spectrum Disorders Panel: **81442**
- Short QT Syndrome Panel: **81403, 81406, 81479 x2**
- Thoracic Aortic Aneurysm Panel: **81410**
- *TTN* Full Mutation Analysis: **81479**
- Targeted variant analysis and deletion/duplication: **call for information.**

Please call 1-866-450-4198 for current pricing, insurance preauthorization or with any billing questions.

Shipping Instructions:

Please enclose test requisition with sample. All information must be completed before sample can be processed. Place samples in styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Saturday.

Ship to:

Genetics and Genomics Diagnostic Laboratory
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474

Note: Cardiovascular diseases panel cases with negative or uncertain findings can be reflexed to Whole Exome Sequencing (WES). A separate test order and a signed consent form is required for all WES testing. In addition, including biological parental samples is strongly encouraged to assist with the analysis of WES and to increase test yield. Reflex to WES orders can either be placed simultaneously or separately. Separate reflex to WES orders are subject to review prior to the initiation of testing. Please see our website at www.cincinnatichildrens.org/exome to obtain a WES test requisition and consent form.

Arrhythmia Disorders:

Panel Name	# of Genes	Genes
Comprehensive Arrhythmia Panel	81	*ABCC9, ACTN2, AKAP9, ANK2, ANKRD1, BAG3, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CPT1A, CTNNA3, DES, DSC2, DSG2, DSP, EMD, FLNC, GATA4, GATA5, GATA6, GJA5, GNB5, GPD1L, GYG1, HCN4, JUP, KCNA5, KCND3, KCNE1, KCNE1L, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK3, KCNQ1, KCNQ3, LDB3, LMNA, MYH6, MYH7, MYL4, NKX2-5, NOS1AP, NPPA, NUP155, PDLIM3, PKP2, PLN, PPA2, PRKAG2, RANGRF, RBM20, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SLC25A20, SLMAP, SNTA1, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TNNI3K, TNNT2, TRDN, TRPM4, TTN*
Atrial Fibrillation (A Fib) Panel	28	*ABCC9, CACNB2, GATA4, GATA6, GJA5, HCN4, KCNA5, KCND3, KCNE1, KCNE1L, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LDB3, LMNA, NPPA, NUP155, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, TBX5*
Atrioventricular Block (AV Block) Panel	7	*DES, EMD, LMNA, NKX2-5, SCN1B, SCN5A, TRPM4*
Brugada Syndrome Panel	21	*ABCC9, CACNA1C, CACNA2D1, CACNB2, CAV3, GPD1L, HCN4, KCND3, KCNE1L, KCNE3, KCNH2, KCNJ8, PKP2, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, SLMAP, TRPM4*
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel	11	*ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, KCNQ1, RYR2, SCN5A, TECRL, TRDN*
Long QT Syndrome Panel	19	*AKAP9, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, NOS1AP, SCN4B, SCN5A, SNTA1, TECRL, TRDN*
Short QT Syndrome Panel	6	*CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1*

Connective Tissue Disorders:

Panel Name	# of Genes	Genes
Thoracic Aortic Aneurysm Panel	49	*ABL1, ACTA2, ADAMTS10, ADAMTS2, ADAMTSL4, ALDH18A1, BGN, CBS, CHST14, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GATA5, LOX, LTBP3, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TAB2, TGFB2, TGFB3, TGFBR1, TGFBR2, TGFBR3, TNXB, ZNF469*
Marfan Syndrome and MFS Related Disorders Panel	3	*FBN1, TGFBR1, TGFBR2*

Cardiomyopathy Disorders:

Panel Name	# of Genes	Genes
Comprehensive Cardiomyopathy Panel	135	*AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALMS1, ALPK3, ANKRD1, BAG3, BRAF, CACNA1C, CALR3, CASQ2, CAV3, CAVIN4, CBL, CDH2, CHRM2, COX15, CPT2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, ELAC2, EMD, EPG5, EYA4, FBXO32, FHL1, FHL2, FKRP, FKTN, FLNC, FXN, GAA, GATA4, GATA6, GATAD1, GBE1, GLA, GLB1, HAND1, HCN4, HRAS, ILK, JPH2, JUP, KRAS, LAMA4, LAMP2, LDB3, LMNA, LRRC10, LZTR1, MAP2K1, MAP2K2, MIB1, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NDUFAF2, NEBL, NEXN, NF1, NKX2-5, NPPA, NRAS, PCCA, PCCB, PDLIM3, PKP2, PLEKHM2, PLN, PPP1CB, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RBM20, RIT1, RMND1, RYR2, SCN5A, SCO2, SDHA, SGCD, SHOC2, SLC22A5, SLC25A4, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE2, TAB2, TAZ, TBX20, TBX5, TCAP, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TTN, TTR, TXNRD2, VCL*
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel	18	*CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYH7, PKP2, PLN, RYR2, SCN5A, TGFB3, TMEM43, TTN*

Cardiomyopathy Disorders (continued):

Panel Name	# of Genes	Genes
Dilated Cardiomyopathy (DCM) Panel	73	*ABCC9, ACTC1, ACTN2, ALMS1, ALPK3, ANKRD1, BAG3, CAV3, CHRM2, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL2, FKRP, FKTN, FLNC, GATA6, GA-TAD1, GLA, HCN4, ILK, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEBL, NEXN, NKX2-5, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RYR2, SCN5A, SDHA, SGCD, SPEG, TAZ, TBX20, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TTN, TTR, TXNRD2, VCL*
Hypertrophic Cardiomyopathy (HCM) Panel	98	*AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALPK3, ANKRD1, BAG3, BRAF, CACNA1C, CALR3, CAV3, CBL, COX15, CPT2, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, DTNA, ELAC2, EMD, EPG5, FHL1, FKTN, FLNC, FXN, GAA, GATA4, GATAD1, GLA, HRAS, ILK, JPH2, JUP, KRAS, LAMA4, LAMP2, LDB3, LMNA, LZTR1, MAP2K1, MAP2K2, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NDUFAF2, NEBL, NEXN, NF1, NRAS, PDLIM3, PKP2, PLN, PPP1CB, PRKAG2, PTPN11, RAF1, RBM20, RIT1, RYR2, SCN5A, SCO2, SDHA, SGCD, SHOC2, SLC22A5, SLC25A4, SOS1, SOS2, SPRED1, SURF1, TAZ, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL*
Left Ventricular Noncompaction (LVNC) Panel	36	*ABCC9, ACTC1, ACTN2, BAG3, CTNNA3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, FBXO32, FLNC, HCN4, JPH2, JUP, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, PKP2, PLEKHM2, PLN, RAF1, RBM20, RYR2, SCN5A,TAZ, TCAP, TNNT2, TPM1, TTN, VCL*
Restrictive Cardiomyopathy (RCM) Panel	9	*ACTC1, BAG3, CRYAB, DES, MYBPC3, MYH7, TNNI3, TNNT2, TTR*

Congenital Heart Disease Disorders:

Panel Name	# of Genes	Genes
Congenital Heart Disease Panel	186	*ABL1, ACTA2, ACTB, ACTC1, ACTG1, ACVR1, ACVR2B, ACVRL1, ADAMTS10, AK7, ALMS1, ANKS6, ARHGAP31, ARMC4, ATRX, B3GAT3, BBS1, BBS10, BBS2, BCL9L, BCOR, BMPR2, BRAF, C21ORF59, CACNA1C, CBL, CCDC103, CCDC11, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CDK13, CENPF, CFAP300, CHD4, CHD7, CITED2, COL2A1, CREBBP, CRELD1, CYR61, DHCR7, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DSG2, DSP, DTNA, EFTUD2, EIF2AK4, ELN, ENG, EVC, EVC2, FBN1, FBN2, FGFR2, FLNA, FLNB, FOXC1, FOXC2, FOXF1, FOXH1, G6PC3, GAS2L2, GAS8, GATA4, GATA5, GATA6, GDF1, GJA1, GJA5, GLI3, GPC3, HAND1, HES7, HRAS, INVS, JAG1, KCNJ2, KIF7, KRAS, LEFTY2, LMNA, LRRC56, LRRC6, MAP2K1, MAP2K2, MCIDAS, MED13L, MEGF8, MEIS2, MID1, MKKS, MKS1, MMP21, MRE11, MYCN, MYH6, NAT10, NEK8, NF1, NIPBL, NKX2-5, NKX2-6, NME8, NODAL, NOTCH1, NOTCH2, NPHP3, NR2F2, NRAS, NSD1, NTRK3, OFD1, PIH1D3, PIK3R2, PITX2, PKD1L1, PKD2, PPP1CB, PQBP1, PRKD1, PRKG1, PRRX1, PTPN11, RAF1, RAI1, RBM10, RIT1, RSPH1, RSPH3, RSPH4A, RSPH9, SALL4, SCN1B, SCN5A, SEMA3E, SHOC2, SHROOM3, SKI, SMAD2, SMAD6, SOS1, SOS2, SOX2, SOX7, SPAG1, SPEG, TAB2, TBX1, TBX20, TBX3, TBX5, TCAP, TCTN2, TFAP2B, TGDS, TGFB2, TGFBR2, TLL1, TTC25, TWIST1, UBR1, VCL, WDR35, ZFPM2, ZIC3, ZMPSTE24, ZMYND10, ZNF469*
Heterotaxy Panel	113	*ACTC1, ACVR2B, AK7, ALMS1, ANKS6, ARMC4, BBS1, BBS10, BBS2, BCL9L, BCOR, BRAF, C21ORF59, CBL, CCDC103, CCDC11, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFAP300, CHD7, CRELD1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, ELN, EVC, EVC2, FOXF1, FOXH1, GAS2L2, GAS8, GATA4, GATA6, GDF1, GJA1, GPC3, HES7, HRAS, INVS, JAG1, KIF7, KRAS, LEFTY2, LMNA, LRRC56, LRRC6, MAP2K1, MAP2K2, MCIDAS, MED13L, MEGF8, MEIS2, MKS1, MMP21, MRE11, NAT10, NEK8, NF1, NKX2-5, NKX2-6, NME8, NODAL, NOTCH1, NOTCH2, NPHP3, NR2F2, NRAS, NSD1, OFD1, PIH1D3, PKD1L1, PKD2, PQBP1, PRRX1, PTPN11, RAF1, RIT1, RSPH1, RSPH3, RSPH4A, RSPH9, SCN5A, SHOC2, SHROOM3, SMAD2, SOS1, SPAG1, TBX1, TBX5, TCTN2, TTC25, UBR1, WDR35, ZIC3, ZMPSTE24, ZMYND10*
RASopathy/Noonan Spectrum Disorders Panel	31	*A2ML1, ACTB, ACTG1, BRAF, CBL, CDC42, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NF2, NRAS, NSUN2, PPP1CB, PTEN, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1, TBCK, TSC1, TSC2*

Other Panels:

Panel Name	# of Genes	Genes
Pulmonary Arterial Hypertension (PAH) Panel	20	*ACVRL1, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, GDF2, GGCX, KCNA5, KCNK3, KLK1, NFU1, NOTCH1, NOTCH3, RASA1, SMAD4, SMAD9, SOX17, TBX4, TOPBP1*

Note: Single gene sequencing is available for all genes listed in the next-generation panels through custom gene sequencing.

Targeted deletion and duplication analysis is available for all genes except *A2ML1, ABCC9, ABL1, ACTA1, ACTA2, ACTG1, ACTN2, ACVR1, ACVR2B, AKAP9, ALPK3, ANKRD1, APOB (R3500Q/R3500W), ARHGAP31, BCL9L, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CAV1, CAVIN4, CCDC114, CDC42, CDH2, CDK13, CHD4, CHRM2, COL5A2, CRYAB, CSRP3, CTNNA3, CYR61, DSG2, DTNA, FBXO32, FHL2, FOXE3, FOXH1, FXN, GATA5, GA-TAD1, GDF1, GDF2, GNB5, GPD1L, HAND1, HCN4, ILK, JPH2, KCND3, KCNE1, KCNE1L, KCNE2, KCNE3, KCNJ5, KCNJ8, KCNK3, KCNQ3, KLK1, LAMA4, LDB3, LDLR, LEFTY2, LOX, LRRC10, LZTR1, MAP2K1, MAT2A, MCIDAS, MFAP5, MIB1, MID1, MRE11, MYH6, MYL2, MYL3, MYL4, MYLK2, MYOM1, MYPN, NAT10, NEBL, NOS1AP, NOTCH3, NTRK3, PDLIM3, PLEKHM2, PPA2, PPP1CB, PRKD1, PRKG1, PRRX1, PSEN1, PSEN2, RANGRF, RASA2, RBM20, RIT1, RRAS, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SDHA, SHROOM3, SKI, SLMAP, SMAD2, SMAD4, SMAD6, SMAD9, SNTA1, SOS2, SPEG, SYNE2, TCAP, TECRL, TFAP2B, TGFB3, TGFBR3, TLL1, TMEM43, TMPO, TNNC1, TNNI3K, TNNT2, TOPBP1, TRPM4, TTN, TTR, TXNRD2, VCL, WDR35* and *ZMYND10* **at an additional charge.**