Cardiology tests

We combine advanced genetic testing with actionable genetic knowledge to provide the most comprehensive diagnostics for all medical specialties, including inherited cardiovascular diseases.

Blueprint Genetics

Cardiology tests

Genetic diagnostics is becoming a mainstream practice in the field of cardiology and is recommended in guidelines from AHA, HRS-EHRA, ESC, and CCS (PMID: 22075469, 20823110, 21810866, and 21459272). It is also proven to be cost-effective as compared with regular clinical screening (PMID: 22128210 and 21139095).

What genetic diagnostics can offer patients with cardiological diseases

Genetic diagnostics is the most efficient way to subtype hereditary cardiovascular diseases. It forms the basis for selecting the right treatment and making well-informed disease management decisions.

In channelopathies, genetic diagnosis can help to define lifestyle recommendations and select adequate medications, as well as aid in making decisions on placing an ICD. In aortic diseases, identifying the underlying genetic defect can help determine the timing of surgical intervention. For hypertrophic cardiomyopathy (HCM), genetic diagnosis can be the only way to differentiate classical sarcomere disease from phenocopies such as Fabry disease, rasopathies, and glycogen storage disease. Knowing the underlying genetic defect can help to tailor treatment and follow-up strategies that improve a patient's outcome.

As a majority of these diseases are inherited in an autosomal dominant manner and have an elevated risk for sudden cardiac death, genetic diagnosis is considered the most effective tool for family-member risk stratification. Identifying family members at risk makes it possible to begin preventive treatments and/or make lifestyle recommendations, and justify routine follow-ups by health care professionals. Recommendations on lifestyle choices, such as avoiding competitive sports, can significantly benefit individuals carrying channelopathy and cardiomyopathy mutations. Genetic diagnosis can also help in family planning.

Our cardiology tests

We offer comprehensive genetic diagnostics for all hereditary cardiovascular diseases, including diseases such as cardiomyopathies, channelopathies, aortopathies, connective tissue disorders, pulmonary arterial hypertension, congenital heart defects, rasopathies, and inherited dyslipidemias.

Maximized diagnostic potential

- All of our panels include both Sequence and Deletion/Duplication (CNV) Analysis.
- Clinically relevant, deep intronic variants included
- Increased capabilities in difficult-to-sequence regions
- mtDNA analysis included in panels for conditions where symptoms or findings can be caused by mtDNA mutations.
- Panels can be customized by removing or adding up to 200 genes (including the mtDNA genes)

Please visit blueprintgenetics.com/cardiology for more information.

We are continuously developing our services and offering. We may amend service descriptions from time to time by posting new versions on our website. For up-to-date information, please visit blueprintgenetics.com.

Cardiology test panels and genes from Blueprint Genetics

Cardiomyopathy/Heart failure

Panel name, test code, number of genes

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel (21) **Test code: CA0401**	*BAG3* *CDH2* *CTNNA3* *DES* *DSC2*	*DSG2* *DSP* *FLNC JUP* *LDB3* *LEMD2*	*LMNA* *MYH7* *NKX2-5* *PKP2* *PLN*	*RYR2* *SCN5A* *TGFB3* *TMEM43* *TTN*				
mtDNA **Cardiomyopathy Panel (214)** **Test code: CA1201**	*AARS2* *ABCC6* *ABCC9* *ACAD9* *ACADVL* *ACTA1* *ACTC1* *ACTN2* *AGK* *AGL* *ALMS1* *ALPK3* *ANO5* *APOA1* *BAG3* *BRAF* *CALR3* *CAPN3* *CASQ2* *CASZ1* *CBL* *CDH2* *CHRM2* *COX15* *CPT2* *CRYAB* *CSRP3*	*CTNNA3* *DBH* *DES* *DMD* *DNAJC19* *DOLK* *DPM3* *DSC2* *DSG2* *DSP* *DTNA* *DYSF* *EEF1A2* *ELAC2* *EMD* *EPG5* *ETFA* *ETFB* *ETFDH* *FBXL4* *FBXO32* *FHL1* *FHOD3* *FKRP* *FKTN* *FLNC* *FOXD4*	*FOXRED1* *FXN* *GAA* *GATA4* *GATA6* *GATAD1* *GATC* *GBE1* *GFM1* *GLA* *GLB1* *GMPPB* *GSK3B* *GTPBP3* *GUSB* *HADHA* *HAND1* *HCN4* *HFE* *HRAS* *IDUA* *ILK* *ISPD* *JPH2* *JUP* *KLHL24* *KRAS*	*LAMA2* *LAMP2* *LARGE* *LDB3* *LEMD2* *LMNA* *LMOD2* *LRRC10* *LZTR1* *MAP2K1* *MAP2K2* *MAP3K8* *MIPEP* *MLYCD* *MT-ATP6* *MT-ATP8* *MT-CO1* *MT-CO2* *MT-CO3* *MT-CYB* *MT-ND1* *MT-ND2* *MT-ND3* *MT-ND4* *MT-ND4L* *MT-ND5* *MT-ND6*	*MT-RNR1* *MT-RNR2* *MT-TA* *MT-TC* *MT-TD* *MT-TE* *MT-TF* *MT-TG* *MT-TH* *MT-TI* *MT-TK* *MT-TL1* *MT-TL2* *MT-TM* *MT-TN* *MT-TP* *MT-TQ* *MT-TR* *MT-TS1* *MT-TS2* *MT-TT* *MT-TV* *MT-TW* *MT-TY* *MTO1* *MYBPC3* *MYBPHL*	*MYH6* *MYH7* *MYL2* *MYL3* *MYL4* *MYOT* *MYPN* *MYRF* *NDUFAF2* *NEXN* *NF1* *NKX2-5* *NONO* *NRAP* *NRAS* *PCCA* *PCCB* *PKP2* *PLEC* *PLEKHM2* *PLN* *PNPLA2* *PPA2* *PPCS* *PPP1CB* *PRDM16* *PRKAG2*	*PTPN11* *QRSL1* *RAF1* *RASA2* *RBCK1* *RBM20* *RIT1* *RMND1* *RRAS* *RYR2* *SCN5A* *SCNN1B* *SCNN1G* *SCO1* *SCO2* *SDHA* *SELENON* *SGCA* *SGCB* *SGCD* *SGCG* *SHOC2* *SLC22A5* *SLC25A20* *SLC25A4* *SMCHD1* *SOS1*	*SOS2* *SPEG* *SPRED1* *TAB2* *TAZ* *TBX20* *TBX5* *TCAP* *TGFB3* *TMEM43* *TMEM70* *TNNC1* *TNNI3* *TNNI3K* *TNNT2* *TOR1AIP1* *TPM1* *TRIM32* *TSFM* *TTN* *TTR* *VCL* *VCP* *VPS13A* *XK*
mtDNA **Dilated Cardiomyopathy (DCM) Panel (123)** **Test code: CA2201**	*ABCC6* *ABCC9* *ACTA1* *ACTC1* *ACTN2* *ALMS1* *ALPK3* *APOA1* *BAG3* *CASZ1* *CHRM2*	*DES* *DMD* *DOLK* *DPM3* *DSC2* *DSG2* *DSP* *DYSF* *EEF1A2* *EMD* *EPG5*	*ETFA* *ETFB* *ETFDH* *FBXO32* *FHOD3* *FKTN* *FLNC* *FOXD4* *GATA4* *GATA6* *GATC*	*GBE1* *GLB1* *GSK3B* *HAND1* *HCN4* *ILK* *JPH2* *JUP* *KLHL24* *LAMP2* *LDB3*	*LEMD2* *LMNA* *LMOD2* *LRRC10* *MLYCD* *MYBPC3* *MYBPHL* *MYH6* *MYH7* *MYL4* *NKX2-5*	*NRAP* *PCCA* *PCCB* *PKP2* *PLEKHM2* *PLN* *PPCS* *PRDM16* *QRSL1* *RAF1* *RBCK1*	*RBM20* *RMND1* *SCN5A* *SPEG* *TAB2* *TAZ* *TBX20* *TBX5* *TCAP* *TMEM43* *TNNC1*	*TNNI3* *TNNI3K* *TNNT2* *TOR1AIP1* *TPM1* *TTN* *TTR* *VCL* *VPS13A*
mtDNA **Hypertrophic Cardiomyopathy (HCM) Panel (84)** **Test code: CA1901**	*ABCC9* *ACAD9* *ACADVL* *ACTA1* *ACTC1* *ACTN2* *AGK* *AGL* *ALPK3* *APOA1* *BAG3*	*BRAF* *CBL* *COX15* *CSRP3* *ELAC2* *EPG5* *FBXL4* *FHL1* *FHOD3* *FLNC* *FXN*	*GAA* *GLA* *GSK3B* *HRAS* *JPH2* *KLHL24* *LAMP2* *MIPEP* *MT-ATP6* *MT-ATP8* *MT-CO1*	*MT-CO2* *MT-CO3* *MT-CYB* *MT-ND1* *MT-ND2* *MT-ND3* *MT-ND4* *MT-ND4L* *MT-ND5* *MT-ND6* *MT-RNR1*	*MT-RNR2* *MT-TA* *MT-TC* *MT-TD* *MT-TE* *MT-TF* *MT-TG* *MT-TH* *MT-TI* *MT-TK* *MT-TL1*	*MT-TL2* *MT-TM* *MT-TN* *MT-TP* *MT-TQ* *MT-TR* *MT-TS1* *MT-TS2* *MT-TT* *MT-TV* *MT-TW*	*MT-TY* *MYBPC3* *MYH7* *MYL2* *MYL3* *NDUFAF2* *PLN* *PRKAG2* *PTPN11* *RAF1* *RIT1*	*SLC25A4* *SOS1* *TNNC1* *TNNI3* *TNNT2* *TPM1* *TTR*
Left Ventricular Non-Compaction Cardiomyopathy (LVNC) Panel (33) **Test code: CA1801**	*ABCC9* *BAG3* *CTNNA3* *DES* *DMD*	*DSC2* *DSG2* *DSP* *DTNA* *EMD*	*FBXO32* *FLNC* *HCN4* *JPH2* *JUP*	*LAMP2* *LMNA* *MIPEP* *MYBPC3* *MYH6*	*MYH7* *PKP2* *PLEKHM2* *PLN* *RAF1*	*RBM20* *RYR2* *SCN5A* *TCAP* *TNNT2*	*TPM1* *TTN* *VCL*	

Arrhythmia

Panel name, test code, number of genes

Arrhythmia Panel (58) **Test code: CA1601**	*ABCC9* *AKAP9* *ANK2* *BAG3* *CACNA1C* *CACNB2* *CALM1* *CALM2*	*CALM3* *CASQ2* *CAV3* *CDH2* *CTNNA3* *DBH* *DES* *DSC2*	*DSG2* *DSP* *FLNC* *GATA6* *HADHA* *HCN4* *JUP* *KCNA5*	*KCNE1* *KCNE2* *KCNH2* *KCNJ2* *KCNJ5* *KCNQ1* *LDB3* *LEMD2*	*LMNA* *MYH6* *MYH7* *MYL4* *NKX2-5* *NOS1AP* *NUP155* *PKP2*	*PLN* *PPA2* *RYR2* *SALL4* *SCN10A* *SCN1B* *SCN3B* *SCN5A*	*TBX5* *TECRL* *TGFB3* *TMEM43* *TNNI3* *TNNI3K* *TNNT2* *TRDN*	*TRPM4* *TTN*
Atrial Fibrillation Panel (19) **Test code: CA2001**	*CACNB2* *GATA6* *HCN4*	*KCNA5* *KCNE1* *KCNE2*	*KCNH2* *KCNJ2* *KCNJ5*	*KCNQ1* *LDB3* *LMNA*	*NUP155* *RYR2* *SCN10A*	*SCN1B* *SCN3B* *SCN5A*	*TBX5*	
Brugada Syndrome Panel (7) **Test code: CA0901**	*CACNA1C* *CACNB2*	*HCN4* *KCNH2*	*SCN3B* *SCN5A*	*TRPM4*				
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel (10) **Test code: CA0701**	*ANK2* *CALM1*	*CALM2* *CALM3*	*CASQ2* *KCNJ2*	*LMNA* *RYR2*	*TECRL* *TRDN*			
Long QT Syndrome (LQTS) Panel (16) **Test code: CA0301**	*AKAP9* *ANK2*	*CACNA1C* *CALM1*	*CALM2* *CALM3*	*CAV3* *KCNE1*	*KCNE2* *KCNH2*	*KCNJ2* *KCNJ5*	*KCNQ1* *NOS1AP*	*SCN5A* *TECRL*
Short QT Syndrome (SQTS) Panel (5) **Test code: CA2101**	*CACNA1C* *CACNB2*	*KCNH2* *KCNJ2*	*KCNQ1*					

Vascular

Panel name, test code, number of genes

Aorta Panel (52) **Test code: CA1001**	*ABCC6* *ABL1* *ACTA2* *ADAMTS10* *ADAMTS17* *ADAMTS2* *ADAMTSL4*	*ALDH18A1* *ATP7A* *B3GAT3* *BGN* *CBS* *COL1A1* *COL1A2*	*COL2A1* *COL3A1* *COL4A5* *COL5A1* *COL5A2* *COLGALT1* *EFEMP2*	*ELN* *ENPP1* *FBLN5* *FBN1* *FBN2* *FKBP14* *FLNA*	*FOXE3* *GATA5* *HCN4* *LOX* *MAT2A* *MED12* *MFAP5*	*MYH11* *MYLK* *NOTCH1* *PLOD1* *PRKG1* *SKI* *SLC2A10*	*SLC39A13* *SMAD2* *SMAD3* *SMAD4* *SMAD6* *TGFB2* *TGFB3*	*TGFBR1* *TGFBR2* *ZDHHC9*
Ehlers-Danlos Syndrome Panel (32) **Test code: CA0101**	*ABCC6* *ADAMTS2* *ALDH18A1* *ATP6V0A2*	*ATP7A* *B4GALT7* *BGN* *CBS*	*CHST14* *COL11A1* *COL1A1* *COL1A2*	*COL2A1* *COL3A1* *COL5A1* *COL5A2*	*DSE* *EFEMP2* *ELN* *FBLN5*	*FBN1* *FBN2* *FKBP14* *FLNA*	*PLOD1* *PYCR1* *SLC39A13* *SMAD3*	*TGFB2* *TGFBR1* *TGFBR2* *ZNF469*
Marfan Syndrome Panel (34) **Test code: CA0801**	*ABL1* *ADAMTS10* *ADAMTS17* *ADAMTSL4* *B3GAT3*	*BGN* *CBS* *COL11A1* *COL11A2* *COL1A1*	*COL1A2* *COL2A1* *COL3A1* *COL5A1* *COL5A2*	*DLG4* *EFEMP2* *FBN1* *FBN2* *LOX*	*MAT2A* *MED12* *PLOD1* *SKI* *SLC2A10*	*SMAD3* *SMAD6* *TGFB2* *TGFB3* *TGFBR1*	*TGFBR2* *UPF3B* *VCAN* *ZDHHC9*	
Pulmonary Artery Hypertension (PAH) Panel (23) **Test code: CA0601**	*ABCC8* *ACVRL1* *AQP1*	*ATP13A3* *BMPR1B* *BMPR2*	*CAV1* *EIF2AK4* *ENG*	*FOXF1* *GDF2* *KCNA5*	*KCNK3* *KLF2* *NFU1*	*NOTCH3* *RASA1* *SARS2*	*SMAD4* *SMAD9* *SOX17*	*STRA6* *TBX4*

General and congenital

Panel name, test code, number of genes

Comprehensive Cardiology Panel (254) **Test code: CA1301** mtDNA	*AARS2, ABCC6, ABCC9, ACAD9, ACADVL, ACTA1, ACTA2, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, ALMS1, ALPK3, ANK2, ANO5, APOA1, ATPAF2, BAG3, BRAF, CACNA1C, CACNB2, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CASZ1, CAV3, CBL, CDH2, CHRM2, COX15, CPT2, CRYAB, CSRP3, CTNNA3, DBH, DES, DMD, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, DYSF, EEF1A2, ELAC2, EMD, ENPP1, EPG5, ETFA, ETFB, ETFDH, FAH, FBXL4, FBXO32, FHL1, FHOD3, FKRP, FKTN, FLNC, FOXD4, FOXRED1, FXN, GAA, GATA4, GATA5, GATA6, GATAD1, GATC, GBE1, GFM1, GLA, GLB1, GMPPB, GSK3B, GTPBP3, GUSB, HADHA, HAND1, HAND2, HCN4, HFE, HRAS, IDUA, ILK, ISPD, JPH2, JUP, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, KLHL24, KRAS, LAMA2, LAMP2, LARGE, LDB3, LEMD2, LMNA, LMOD2, LRRC10, LZTR1, MAP2K1, MAP2K2, MAP3K8, MIPEP, MLYCD, MRPL3, MRPL44, MRPS22, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTO1, MYBPC3, MYBPHL, MYH6, MYH7, MYL2, MYL3, MYL4, MYO18B, MYOT, MYPN, MYRF, NDUFAF2, NEXN, NF1, NKX2-5, NONO, NOS1AP, NRAP, NRAS, NUP155, PARS2, PCCA, PCCB, PKP2, PLEC, PLEKHM2, PLN, PNPLA2, POMT1, PPA2, PPCS, PPP1CB, PRDM16, PRKAG2, PTPN11, QRSL1, RAF1, RASA2, RBCK1, RBM20, RIT1, RMND1, RRAS, RYR2, SALL4, SCN10A, SCN1B, SCN3B, SCN5A, SCN1B, SCNN1G, SCO1, SCO2, SDHA, SELENON, SGCA, SGCB, SGCD, SGCG, SHOC2, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SMCHD1, SOS1, SOS2, SPEG, SPRED1, STAG2, TAB2, TAZ, TBX20, TBX5, TCAP, TECRL, TGFB3, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM32, TRPM4, TSFM, TTN, TTR, VARS2, VCL, VCP, VPS13A, XK*
Congenital Structural Heart Disease Panel (114) **Test code: CA1501**	*ABL1, ACTA2, ACTB, ACTC1, ACTG1, ACVR1, ACVR2B, ADAMTS10, ADAMTS17, AFF4, AMMECR1, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BMPR2, BRAF, C12ORF57, CBL, CDK13, CDK9, CFAP53, CHD4, CHD7, CHRM2, CREBBP, CRELD1, CTC1, DHCR7, DLL4, DOCK6, EFTUD2, EHMT1, EIF2AK4, ELN, ENG, EOGT, EP300, EVC, EVC2, FLNA, FOXC1, FOXF1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GJA5, GPC3, HAND1, HAND2, HDAC8, HNRNPK, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYNU, LEFTY2, MED12, MED13L, MEIS2, MMP21, MYCN, MYO18B, MYRF, NAA15, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NONO, NOTCH1, NOTCH2, NR2F2, NSD1, PITX2, PKD1L1, PPP1CB, PRDM6, PRKD1, PTPN11, PUF60, RAB23, RAF1, RBM10, RERE, RIT1, SALL4, SMARCB1, SMC1A, SMC3, SOS1, SOS2, STAG2, STRA6, TAB2, TBX1, TBX20, TBX5, TFAP2B, TGDS, TLL1, TMEM94, ZEB2, ZFPM2, ZIC3*
Hereditary Hemorrhagic Telangiectasia (HHT) Panel (6) **Test code: CA0201**	*ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4*
Heterotaxy and Situs Inversus Panel (34) **Test code: CA2301**	*ACVR2B, ANKS6, ARMC4, C21ORF59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CFAP53, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DYX1C1, FOXH1, GDF1, INVS, LEFTY2, LRRC6, MMP21, NODAL, PIH1D3, PITRM1, PKD1L1, SPAG1, TTC25, ZIC3, ZMYND10*
Hyperlipidemia Core Panel (4) **Test code: CA1701**	*APOB, LDLR, LDLRAP1, PCSK9*
Hyperlipidemia Panel (18) **Test code: CA1101**	*ABCA1, ABCG5, ABCG8, ALMS1, APOA1, APOA5, APOB, APOC2, APOC3, APOE, CREB3L3, GPIHBP1, LDLR, LDLRAP1, LIPA, LMF1, LPL, PCSK9*
Liddle Syndrome Panel (2) **Test code: CA1401**	*SCNN1B, SCNN1G*
Noonan Syndrome Panel (35) **Test code: CA0501**	*ACTB, ACTG1, BRAF, CBL, CCNK, CDC42, EPHB4, FGD1, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MAP3K8, MRAS, NF1, NF2, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SASH1, SHOC2, SMARCB1, SOS1, SOS2, SPRED1, STAMBP, SYNGAP1*

Please use the test code when placing orders. We are continuously developing the gene set description and panel composition to match the latest research findings. Please find the most updated list of genes, panels and panel descriptions at: blueprintgenetics.com/cardiology

Ordering a test from us is quick and simple. You can order online using our secure portal, Nucleus.

What we do for you

Changing diagnostics

We use next-generation (NGS) sequencing technology, bringing you fast, affordable, and accurate results.

- Blueprint Genetics utilizes Illumina NGS sequencing technology
- Full coverage of medically actionable genes
- Over 220 tests for all medical specialties, covering over 4,000 genes
- Both sequence and targeted Del/Dup (CNV) analysis
- Whole Exome Sequencing
- Rapid TAT
- Competitive pricing
- Clinical Genetics Support

Providing knowledge

Our interpretation process allows our team of geneticists and clinicians to bring you the most clinically actionable reports available on the market.

- Comprehensive clinical statement prepared by our team of geneticists and clinicians
- Clinical panels with high diagnostic yield
- Easy online ordering and results delivery through our secure portal, Nucleus

We provide comprehensive, actionable genetic diagnostics.

Blueprint Genetics

Find out how to order or contact our customer support at blueprintgenetics.com with any questions about our services. We're here to help!

Join the conversation
#GeneticKnowledge

www.blueprintgenetics.com