Genomic profiling for cardiac disease

Developed in collaboration with leading cardiologists from Washington University School of Medicine

Next-generation sequencing of key genes implicated in arrhythmias and cardiomyopathies for efficient, cost-effective and robust germline variant detection

Concise, expert interpretations by board-certified clinical genomicists, reported back to the ordering physician

Personal support for physicians in result interpretation by our highly-trained genomicists and genetic counselor

Testing covered by most insurance; preauthorization performed by GPS

Clinical utility

The Washington University CardioGene Set is designed to identify causative genetic variation within patients affected with cardiomyopathy or arrhythmia syndromes.

The identification of a pathogenic genetic alteration may be used to solidify a diagnosis, allowing for more appropriate patient management and surveillance. It also enables tailored genetic counseling in both the patient and at-risk family members.

Targeted next-generation sequencing provides clinicians with a powerful tool to enrich patient care and enables the analysis of multiple genes with clinical relevance to enhance the diagnostic yield of testing. Such analysis opens the potential for disease specific therapies over time and may enable genotype-phenotype correlations to be drawn.

Orderable gene sets include:

Arrhythmia (35 genes)
- Brugada (18 genes)
- CPVT (8 genes)
- LQTS (18 genes)
- SQTS (6 genes)

Cardiomyopathy (115 genes)
- ARVC (11 genes)
- DCM (67 genes)
- HCM (62 genes)
- LVNC (15 genes)

Thoracic Aortic Aneurysm and Dissection Gene Set (17 genes)
Indications for testing

Indications for testing include:

- Brugada syndrome
- Catecholaminergic polymorphic ventricular tachycardia (CPVT)
- Long QT syndrome (LQTS)
- Short QT syndrome (SQTS)
- Arrhythmogenic right ventricular cardiomyopathy (ARVC)
- Dilated cardiomyopathy (DCM)
- Hypertrophic cardiomyopathy (HCM)
- Noonan syndrome
- Left ventricular noncompaction (LVNC)
- Aortic aneurysm or aortic dissection

Genes tested

Arrhythmia Gene Set (35 genes): Includes all genes from the following four gene sets.

- Brugada Gene Set (18 genes): CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCNA3, KCNE3, KCNE5 (KCNE1L), KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A5, SLMAP, TRPM4
- CPVT Gene Set (8 genes): ANK2, CALM1, CALM2, CASQ2, KCNJ2, RYR2, TRDN
- LQTS Gene Set (6 genes): AKAP9, ANK2, CACNA1C, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ5, KCNO1, SCN4B, SCN5A, SNTA1, TRDN
- SQTS Gene Set (6 genes): CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNO1

Cardiomyopathy Gene Set (115 genes): Includes all genes from the following four gene sets.

- ARVC Gene Set (11 genes): CTNNA3, DES, DSC2, DSG2, DSP, JUP, PDLIM3, PKP2, RYR1, RYR2, TMEM43
- DCM Gene Set (67 genes): ABC9, ACADVL, ACTA1, ACTC1, ACTN2, ALM1S1, ALPK3, ANKRD1, BAG3, CBL, CHRM2, CPT2, CRYPAB, CSR3P, CTF1, DES, DMD, DNAJC19, DOLK, EDM, EYA4, FHL1, FHL2, FKN, FLNC, GATA4, GATA6, GATA1, HFE, ILK, LAM2, LDB3, LMNA, LRRC10, MURC, (C4VNA), MYBPC3, MYH6, MYH7, MYPN, NEB1, NEXN, NKX2-5, NPPA, PPLIM3, PLEKH2, PLN, RBNM, SCN5A, SMDA, SEPN1, SELENON, SGCD, SGCG, SLC22A5, TAZ, TBX20, TCA, TEM7M0, TMN1C, TNNS3, TNNT2, TPM1, TTN, TXNRD2, VCL, ZBTB17
- HCM Gene Set (62 genes): A2ML1, ACADVL, ACTA1, ACTC1, ACTN2, AGL, BRAF, CALR3, CBL, CPT2, CRYPAB, CSR3P, ELAC2, FLNC, GA, GAT4, GLA, HFE, HRA, JPH2, KRTS, LAM2, MAP2K1, MAP2K2, MT1, MYBLC3, MYH6, MYH7, MYL2, MYL3, MYL2, MYOM1, MYOZ2, MYOZ2, NPPA, NPPB, NPPC, RAS, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RASA1, RIT1, RRAS, SEPN1 (SELENON), SGCD, SGCG, SLC22A5, TAZ, TBX20, TCA, TEM7M0, TNN1C, TNNS3, TNNT2, TPM1, TRIM63, TTR, ZBTB17
- LVNC Gene Set (15 genes): ACTC1, CASQ2, DNAC19, DNTA, LAMA4, LDB3, LMNA, MYH1, MYB3, MYH7, PRDM16, TAZ, TNNT2, VCL, YWHAE

Thoracic Aortic Aneurysm and Dissection (TAAAD) Gene Set (17 genes): ACTA2, COL3A1, COL5A2, EFEMP2, FN1, FN2, LOX, MYH11, MYLK, NOTCH1, SL2A10, SMAD3, SMAD4, TGFB2, TGFB3, TGFB1R1, TGFB2R

Testing methodology

Tests are performed using targeted hybridization capture coupled with next-generation sequencing (NGS) in our CAP/CLIA labs for comprehensive coverage of all coding exons of ordered genes. Types of variation detected include single nucleotide variants (SNVs) and small insertions and deletions (indels).

Results and interpretation

DNA sequence data are analyzed by GPS’s clinically validated bioinformatics pipeline to identify and annotate genetic variants associated with arrhythmias or cardiomyopathies. Variants are interpreted by a board-certified clinical genomicsist in the context of the patient’s disease. Those that are most likely to account for the observed clinical phenotype based on evidence from the medical literature are highlighted. Results are returned to the ordering physician in a concise report.

Specimen requirements

Specimen types accepted include 2-5 mL of peripheral blood in a lavender-top EDTA tube. Specimen kits are available upon request.

Turnaround time

The turnaround time for testing and interpretation is four weeks after a specimen arrives following pre-authorization.

Ordering

To order a test, submit a completed requisition form (available on our website) by fax or email. Tests are reimbursable, and GPS performs insurance preauthorization.

Selected references


Contact us to order a test or for more info

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