Comprehensive Cardiomyopathy Panel

Test Name:
Comprehensive Cardiomyopathy Panel

Alias:

Cardiomyopathy
Hereditary Cardiomyopathy
DCM
Dilated Cardiomyopathy
HCM
Hypertrophic Cardiomyopathy
Left ventricular noncompaction
LVNC
Next-Generation Sequencing
NextGen Sequencing Test

Overview / Description of the Test:

This test utilizes next-generation sequencing to detect single nucleotide variants, insertions and deletions up to 20 bp, and copy number variants in 86 genes associated with hereditary forms of cardiomyopathy.

Identification of a disease-causing variant may assist with diagnosis, prognosis, clinical management, familial screening, and genetic counseling for hereditary forms of cardiomyopathy; including hypertrophic cardiomyopathy and dilated cardiomyopathy.

List of Gene Targets:

ABCC9, ACAD9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, BAG3, BMP10, BRAF, CDH2, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, ELAC2, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GLA, HCN4, HRAS, JPH2, JUP, KRAS, LAMP2, LMNA, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, MYLK3, MYPN, NEXN, NKX2-5, NRAS, PCCA, PCCB, PKP2, PLN, PPA2, PPCS, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RIT1, RYR2, SCN5A, SGCD, SHOC2, SLC22A5, SOS1, SOS2, SYNE2, TAFAZZIN, TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TTN, TTR, and VCL

Method Name:

Sequence capture and targeted next-generation sequencing

Specimen Type(s):

Whole blood collected in K2 EDTA; Preferred: Lavender top (BD Vacutainer EDTA, T944), Acceptable: BD Vacutainer EDTA tube, 4mL (13x75 mm), 6mL (13x100 mm), and 10mL (16x100 mm)

Shipping Instructions:

Specimens preferred to arrive at Helix within 96 hours of collection at room temperature.

Estimated Turnaround Time:

7-24 days

Performing Laboratory Information:

Location: 10170 Sorrento Valley Road, Suite 100, San Diego, CA 92121 (CLIA# 05D2117342, CAP #9382893, NYS PFI: 9396)
Director: Cotter, Philip D. Ph.D., FACMG, FFSc (RCPA)
Hours of Operation: Monday-Sunday (6AM-10PM PST)