Hypertrophic Cardiomyopathy Panel

Test Name:

Hypertrophic Cardiomyopathy Panel

Alias:

Hypertrophic cardiomyopathy
HCM

Overview / Description of the Test:

This test is intended for genetic screening for and diagnosis of hypertrophic cardiomyopathy.

This test utilizes next-generation sequencing to detect single nucleotide changes, insertion/deletions of up to 20 base pairs, and copy number variants in 48 genes associated with hereditary forms of hypertrophic cardiomyopathy.

Identification of disease-causing variant(s) may assist with the diagnosis, prognosis, clinical management, familial screening, and genetic counseling for hereditary forms of hypertrophic cardiomyopathy.

List of Gene Targets:

ACAD9, ACADVL, ACTC1, ACTN2, AGL, ALPK3, BRAF, CPT2, CSRP3, ELAC2, FHL1, FLNC, GAA, GLA, HRAS, JPH2, KRAS, LAMP2, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, NEXN, NRAS, PLN, PPA2, PRKAG2, PTPN11, RAF1, RIT1, SHOC2, SLC22A5, SOS1, SOS2, TCAP, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, 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)