GenCardio Cardiomyopathy Panel

GENOME EXPLORATIONS
A DIVISION OF COMPASS LABORATORY SERVICES LLC

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Introduction

The GenCardio Cardiomyopathy Panel is a collection of multiplexed PCR primer assays for targeted enrichment of the coding (exonic) regions of 58 genes mutated in hereditary cardiomyopathy. The two major forms of primary cardiomyopathy are hypertrophic and dilated. Hypertrophic cardiomyopathy commonly presents with left ventricular hypertrophy, often due to high blood pressure during the diastolic (resting) phase of the heartbeat. Dilated cardiomyopathy commonly presents with dilated ventricles, often due to increased systolic blood pressure. There is a significant genetic component to both of these forms of cardiomyopathy, including 50% of hypertrophic patients and 20–35% of dilated cardiomyopathy patients. There are additional rarer forms of familial cardiomyopathy, including some inherited diseases with broader phenotypes that include cardiomyopathy. Although many pathogenic mutations for hereditary cardiomyopathy have been discovered, sequencing of the full coding region of each related gene allows insight into potential novel pathogenic mutations. This panel allows researchers to comprehensively study genes involved in hereditary cardiomyopathy, and was developed using a variety of resources such as OMIM® (Online Mendelian Inheritance in Man), Clinvar, and dbSNP.

GenCardio Cardiomyopathy Panel Genes

Hypertrophic Cardiomyopathy:
ACTC1, ACTN2, CALR3, CAV3, COX15, CRYAB, CSRP3, JPH2, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, NEXN, PLN, PRKAG2, SCO2, SLC25A4, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL

Dilated Cardiomyopathy:
ABCC9, ACTC1, ACTN2, BAG3, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, EMD, EYA4, FKTN, GATAD1, JUP (CTNNG), LAMA4, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYPN, NEXN, PLN, PSEN1, PSEN2, RBM20, SCN5A, SDHA, SGCD, TAZ, TBX20, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL

Restrictive Cardiomyopathy:
ACTC1, DES, MYH7, TNNI3, TNNT2

Arrhythmogenic Cardiomyopathy:
DSC2, DSG2, DSP, JUP (CTNNG), PKP2, RYR2, TGFB3, TMEM43

LV Noncompaction:
ACTC1, DTNA, LDB3, LMNA, MYBPC3, MYH7, TAZ

Inherited Diseases:
Fabry Disease: GLA
Pompe Disease: GAA
Danon Disease: LAMP2
Barth Syndrome: TAZ
DMD-Associated Dilated Cardiomyopathy: DMD
Salih Myopathy: TTN