Hypertrophic Cardiomyopathy: Deletion/Duplication Panel

Test Code: MD091  
Turnaround time: 2 weeks  
CPT Codes: 81228 x1, 81479 x1

Condition Description

Hereditary hypertrophic cardiomyopathy (HCM) is inherited in an autosomal dominant manner. HCM is characterized by left ventricular hypertrophy in the absence of a predisposing cardiac or cardiovascular condition. The manifestation of HCM is extremely variable, even within the same family, and can range from asymptomatic to progressive heart failure. Other features include syncope, presyncope, shortness of breath, chest pain, orthostasis, and palpitations. The onset of HCM is usually during adolescence or young adulthood; however, it can range from infancy to much later in adult life. The prevalence of HCM is approximately 1 in 500 and ~55-70% of cases are caused by a mutation in one of the genes that encode a part of the sarcomere.

Reference:  
- GeneReviews

Genes

ACTC1, CAV3, GLA, JPH2, LAMP2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYQ2, MYPN, PRKAG2, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR

Indications

This test is indicated for individuals with:

- Confirmation of a clinical diagnosis of hereditary hypertrophic cardiomyopathy (HCM).
- Carrier testing in adults with a family history of hereditary hypertrophic cardiomyopathy (HCM).

Methodology

For Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

Detection

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood

Specimen Requirements:

In EDTA (purple top) tube:  
- Infants (2 years): 3-5 ml  
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

Type: Isolated DNA

Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.
Related Tests

- Comprehensive Cardiomyopathy Sequencing Panel.
- Hypertrophic Cardiomyopathy: Sequencing Panel.