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, MYOZ2, 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.

### 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 (EDTA)**

**Specimen Requirements:**

EDTA (Purple Top)  
Infants and Young Children (2 years of age to 10 years old): 3-5 ml  
Older Children & Adults: 5-10 ml  
Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

**Type: DNA, Isolated**

**Specimen Requirements:**

Microtainer 3µg  
Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

### Related Tests

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

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