The MYBPC3 gene encodes cardiac myosin-binding protein C (MYBPC3) which is arrayed transversely in sarcomere A-bands and binds myosin heavy chain in thick filaments and titin in elastic filaments. The MYBPC3 gene contains 35 exons and spans around 21 kb genomic distance that was mapped to chromosome 11p11.2. Defects in this gene are the cause of dilated and familial hypertrophic cardiomyopathy (DCM and FHCM). DCM is a type of cardiomyopathy in which the heart becomes weakened and enlarged and cannot pump blood efficiently. About 25–35% of patients have familial forms of the disease. The disease is genetically heterogeneous, but the most common form of its transmission is an autosomal dominant pattern. FHCM is an autosomal dominant inherited disease characterized by a portion of the myocardium is hypertrophied (thickened). MYBPC3 gene is also associated with left ventricular noncompaction (LVNC), which is rare congenital cardiomyopathy which results from the failure of myocardial development during embryogenesis. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for MYBPC3 mutations. Individuals are tested by DNA sequencing of the coding exons of the MYBPC3 gene. We strongly recommend initial testing of a clearly affected individual, if available, in order to provide the greatest test sensitivity and clearest interpretation of results for subsequent family members. Genetic counseling is recommended for all individuals.

**METHODOLOGY**

Genomic DNA is analyzed for MYBPC3 mutations by DNA sequencing of the coding exons of the MYBPC3 gene, as well as the exon/intron junctions and a portion of the 5’ and 3’ untranslated regions. Patient DNA is sequenced in both the forward and reverse orientations. If a mutation is identified, additional family members are analyzed only for the familial mutation by automatic fluorescent DNA sequencing.

**SERVICE FEES**

<table>
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<tr>
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<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tr>
<td>Index Case (Male or Female)</td>
<td>$1500 per sample</td>
<td>81407</td>
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<tr>
<td>Additional Family Members</td>
<td>$300 per sample; Known familial mutation only</td>
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**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons of MYBPC3.

**SPECIMEN REQUIREMENTS**

- **Blood (preferred):** EDTA (purple-top) tubes: 
  - Adult: 5 cc
  - Child: 5 cc
  - Infant: 2-3 cc
- **Tissue:** Frozen (preferred), RNAlater
- **Other Body Fluids and Formalin-fixed, Paraffin-embedded Tissue:** Call to inquire