Myosins are a large family of motor proteins that share the common features of ATP hydrolysis, actin binding and potential for kinetic energy transduction. Myosin light chain-2 (MYL2) gene encodes the regulatory light chain associated with cardiac myosin beta (or slow) heavy chain. MYL2 gene contains 7 coding exons and spans a around 9.9 kb genomic distance which was mapped to chromosome 12q24.11. Mutations in this gene are associated with familial hypertrophic cardiomyopathy 10 and MYL2-related familial hypertrophic cardiomyopathy. MYL2 mutations demonstrate autosomal dominant inheritance with a broad range of clinical severity both within and between families. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for MYL2 mutations. Individuals are tested by DNA sequencing of the coding exons of the MYL2 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.

**REASONS FOR REFERRAL**

Molecular confirmation of the diagnosis of familial hypertrophic cardiomyopathy 10 and MYL2-related familial hypertrophic cardiomyopathy.

**METHODOLOGY**

Genomic DNA is analyzed for MYL2 mutations by DNA sequencing of the coding exons of the MYL2 gene, as well as the exon/intron junctions and a portion of the 5’ and 3’ untranslated region. 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>
<thead>
<tr>
<th>Service</th>
<th>CPT Codes</th>
<th>Direct and Institutional Billing</th>
</tr>
</thead>
<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>81405</td>
<td>$600 per sample</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>81403</td>
<td>$300 per sample; known familial mutation only</td>
</tr>
</tbody>
</table>

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-7 of MYL2.

**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