Myogenic factors (MYF) belong to the basic helix-loopelix (bHLH) transcription factor family and are expressed solely in skeletal muscle lineages. The human *MYF6* gene encompasses six exons and encodes a protein of 729 amino acids. *MYF6* gene has been mapped to chromosome 12q21.31. MYFs determine specification and differentiation of skeletal muscle cells. *MYF6* mutations are associated with myopathy and severe course of Becker muscular dystrophy (BMD). Cardiac involvement is a frequent feature of BMD. Cardiomyopathy occurs in BMD patients and is the main factor influencing survival. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for *MYF6* mutations. Individuals are tested by automatic fluorescent DNA sequencing of the coding exons of the *MYF6* 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 cardiomyopathy.

## METHODOLOGY

Genomic DNA is analyzed for *MYF6* mutations by automatic fluorescent DNA sequencing of the coding exons of the *MYF6* 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></th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<td>Index Case (Male or Female)</td>
<td>$600 per sample</td>
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<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
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## SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-6 of *MYF6*.

## 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 or Formalin-fixed, Paraffin-embedded Tissue:** Call to inquire