ALMS1 (Alstrom syndrome 1) encodes a protein containing a large tandem-repeat domain. The human gene ALMS1 spans 224 kb of genomic DNA and encodes a 4168 amino acid. It is located at 2p13.1. Mutations in this gene have been associated with Alstrom syndrome. Alström syndrome is characterized by a typically transient dilating cardiomyopathy in infancy, suggesting that mitogenic cardiomyopathy represents the extreme phenotype, resulting in demise before the other clinical symptoms become evident. Cardiomyopathy is a primary heart muscle disorder caused by functional abnormalities in cardiomyocytes and a major cause of cardiac sudden death and progressive heart failure. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for ALMS1 mutations. Individuals are tested by automatic fluorescent DNA sequencing of the coding exons of the ALMS1 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 ALMS1 mutations by automatic fluorescent DNA sequencing of the coding exons of the ALMS1 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>Index Case (Male or Female)</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>$1,100 per sample</td>
<td>81406</td>
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</table>

| Additional Family Members   | $300 per sample; known familial mutation only | 81403 |

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-23 of ALMS1.

**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