GATAD1 encodes GATA zinc finger domain-containing protein 1, which binds to a histone modification site that regulates gene expression. The human GATAD1 gene contains 5 coding exons that span about 9 kb located at 7q21.2. Dilated cardiomyopathy (DCM) is a heritable, genetically heterogeneous disorder that typically exhibits autosomal dominant inheritance. However, mutations in GATAD have been associated with autosomal recessive dilated cardiomyopathy. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for GATAD1 mutations. Individuals are tested by DNA sequencing of the coding exons of the GATAD1 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 dilated cardiomyopathy.

**METHODOLOGY**
Genomic DNA is analyzed for GATAD1 mutations by automatic fluorescent DNA sequencing of the coding exons of the GATAD1 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>Description</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>$500 per sample</td>
<td>81404</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
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</tbody>
</table>

**SENSITIVITY**
DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-5 of GATAD1.

**SPECIMEN REQUIREMENTS**

**Blood (preferred):** EDTA (purple-top) tubes:  
- **Adult:** 5 cc  
- **Child:** 5 cc  
- **Infant:** 2-3 cc

**Tissue:** Frozen (preferred) or RNAlater

**Other Body Fluids or Formalin-fixed, Paraffin-embedded Tissue:** Call to inquire