The **CALM2** gene encodes calmodulin 2, which is one of the family of calcium-modulated proteins of which nearly 20 members have been found. They are identified by their occurrence in the cytosol or on membranes facing the cytosol and by a high affinity for calcium. Its functions include roles in growth and the cell cycle as well as in signal transduction and the synthesis and release of neurotransmitters. The **CALM2** gene contains 6 exons and spans more than 16 kb that were mapped to chromosome 2p21.3-p21.1. Mutations in this gene are associated with cardiac arrhythmias such as catecholaminergic polymorphic ventricular tachycardia (CPVT) and long QT syndrome (LQT). Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for **CALM2** mutations. Individuals are tested by DNA sequencing of the coding exons of the **CALM2** 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 catecholaminergic polymorphic ventricular tachycardia (CPVT) and long QT syndrome (LQT).

### METHODOLOGY

Genomic DNA is analyzed for **CALM2** mutations by DNA sequencing of the coding exons of the **CALM2** 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>
<thead>
<tr>
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<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tbody>
<tr>
<td><strong>Index Case (Male or Female)</strong></td>
<td>$600 per sample</td>
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<tr>
<td><strong>Additional Family Members</strong></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 of **CALM2**.

### 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