The KCNE1L gene contains one exon and spans around 1.5 kb mapped to Xq22.3. KCNE1L (KCNE1-like) encodes a membrane protein that has 56% sequence similarity to the potassium channel KCNE1 gene product, a member of the isk-related voltage-gated subfamily of potassium channels. The protein contains a single transmembrane domain surrounded by many charge residues and 2 potential N-glycosylation sites. Mutation in this gene may be involved in the cardiac and neurologic abnormalities found in AMME contiguous gene syndrome and familial atrial fibrillation. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for KCNE1L mutations. Individuals are tested by DNA sequencing of the coding exons of the KCNE1L 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 BrS, cardiac and neurologic abnormalities found in AMME contiguous gene syndrome and familial atrial fibrillation.

**METHODOLOGY**

Genomic DNA is analyzed for KCNE1L mutations by DNA sequencing of the one coding exon of the KCNE1L 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>
<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>81403</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
</tr>
</tbody>
</table>

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exon 1 of KCNE1L.

**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

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