KCNH2 gene, also known as HERG, encodes the pore-forming subunit of a rapidly activating–delayed rectifier potassium channel that plays an essential role in the final repolarization of the ventricular action potential. KCNH2 gene contains 15 exons spanning approximately 19 kb of genomic DNA that was mapped to chromosome 7q35-q36 by fluorescence in situ hybridization. Multiple mutations in KCNH2 gene have been identified to be associated with long-QT syndrome type 2 (LQT2) and short-QT syndrome 1 (SQT1). KCNH2 mutations demonstrate autosomal dominant inheritance with a broad range of clinical severity both within and between families. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for KCNH2 mutations. Individuals are tested by DNA sequencing of the coding exons of the KCNH2 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 long-QT syndrome type 2 (LQT2) and short-QT syndrome 1 (SQT1).

**METHODOLOGY**

Genomic DNA is analyzed for KCNH2 mutations by DNA sequencing of the coding exons of the KCNH2 gene, as well as the exon/intron junctions and a portion of the 3’ and 5’ 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>$1,000 per sample</td>
<td>81406</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
</tr>
</tbody>
</table>

**SENSITIVITY**


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

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