SNTA1 (syntrophin, alpha 1) is a member of the syntrophin gene family which contains at least two other structurally-related genes. SNTA1 gene contains eight exons spanning 35.9 kb of genomic distance that was mapped to chromosome 20q11.2. Human SNTA1 encodes a deduced 505-amino acid peripheral membrane protein associated with dystrophin and dystrophin-related proteins which is missing in patients with Duchenne Muscular Dystrophy and is present in reduced amounts in patients with Becker Muscular Dystrophy. Multiple mutations in SNTA1 have been identified in patients with long-QT syndrome type 12 (LQT12). SNTA1 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 SNTA1 mutations. Individuals are tested by DNA sequencing of the coding exons of the SNTA1 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 12 (LQT12).

**METHODOLOGY**

Genomic DNA is analyzed for SNTA1 mutations by DNA sequencing of the coding exons of the SNTA1 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>
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<tr>
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<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<td>Index Case (Male or Female)</td>
<td>$750 per sample</td>
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<tr>
<td>Additional Family Members</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 1-8 of SNTA1.

**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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