**SGCG MUTATION ANALYSIS**

JOHN WELSH CARDIOVASCULAR
DIAGNOSTIC LABORATORY

SGCG (SARCOGLYCAN, GAMMA) gene encodes gamma-sarcoglycan, one of several sarcolemmal transmembrane glycoproteins that interact with dystrophin, which is a 291 amino acids peptide with a molecular mass of 32.4 kD. SGCG located at 13q12.12 and encompasses 8 exons. The dystrophin-glycoprotein complex (DGC) spans the sarcolemma and is comprised of dystrophin, syntrophin, alpha- and beta-dystroglycans and sarcoglycans. The DGC provides a structural link between the subsarcolemmal cytoskeleton and the extracellular matrix of muscle cells. Defects in the encoded protein can lead to early onset autosomal recessive muscular dystrophy, in particular limb-girdle muscular dystrophy, type 2C (LGMD2C). LGMD2C are muscular dystrophies in which the development of a dilated cardiomyopathy is common. Arrhythmias and conduction disease occur after the development of the dilated cardiomyopathy. Patients with SGCG mutations usually exhibit muscular dystrophies which affect skeletal muscle and cardiac muscle. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for SGCG mutations. Individuals are tested by automatic fluorescent DNA sequencing of the coding exons of the SGCG 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 atrial arrhythmias and ventricular arrhythmias.

**METHODOLOGY**

Genomic DNA is analyzed for SGCG mutations by DNA sequencing of the coding exons of the SGCG 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>Index Case (Male or Female)</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<td>$800 per sample</td>
<td>81405</td>
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<th>Additional Family Members</th>
<th>$300 per sample; known familial mutation only</th>
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**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 2-8 of SGCG.

**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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[Contact Information]