GJA5 MUTATION ANALYSIS
JOHN WELSH CARDIOVASCULAR
DIAGNOSTIC LABORATORY

The GJA5 gene encodes connexin 40 which oligomerize to form intercellular channels, called gap junctions, through which ions and small molecules move between adjacent cells. The GJA5 gene contains 3 exons and spans about 25 kb that were mapped to chromosome 1q21.1. Mutations in this gene are associated with atrial fibrillation (AF). Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for GJA5 mutations. Individuals are tested by DNA sequencing of the coding exons of the GJA5 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 CHD and familial atrial fibrillation (FAF).

METHODOLOGY

Genomic DNA is analyzed for GJA5 mutations by DNA sequencing of the coding exons of the GJA5 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>Service Description</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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</thead>
<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>$500 per sample</td>
<td>81403</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 of GJA5.

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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