**EYA1 MUTATION ANALYSIS**

**JOHN WELSH CARDIOVASCULAR DIAGNOSTIC LABORATORY**

**EYA1 (Eyes Absent 1)** encodes a member of the EYA family protein phosphatase, which is required for regulating genes encoding growth control and signaling molecules, modulating precursor cell proliferation. Human EYA1 gene contains 18 exons which have been mapped to chromosome 8q13.3. Mutations in the EYA1 gene can cause deletions of chromosome 22q11.2, which leads to human del22q11 syndrome, DiGeorge syndrome (DGS), and velo-cardio-facial Syndrome (VCFS). Affected patients exhibit a wide spectrum of developmental defects, including craniofacial anomalies, dysmorphogenesis of cardiovascular structures, and hypoplasia of the thymus and parathyroid glands. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for EYA1 mutations. Individuals are tested by automatic fluorescent DNA sequencing of the coding exons of the EYA1 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, the del22q11 syndrome, DiGeorge syndrome (DGS), and velo-cardio-facial Syndrome (VCFS).

### METHODOLOGY

Genomic DNA is analyzed for EYA1 mutations by DNA sequencing of the coding exons of the EYA1 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>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>$1,150 per sample</td>
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<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
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### SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-18 of EYA1.

### 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