**FOXC2 MUTATION ANALYSIS**

**JOHN WELSH CARDIOVASCULAR DIAGNOSTIC LABORATORY**

*FOXC2* gene belongs to the forkhead family of transcription factors which is characterized by a distinct DNA-binding forkhead domain. *FOXC2* gene contains a single coding exon and spans approximately 1.5 kb and has been mapped to chromosome 16q24.1. *FOXC2* might be involved in the formation of special mesenchymal tissues. Diseases associated with *FOXC2* include lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, and lymphedema-distichiasis syndrome. *FOXC2* 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 *FOXC2* mutations. Individuals are tested by DNA sequencing of the coding exons of the *FOXC2* 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 congenital heart diseases (CHD).

**METHODOLOGY**

Genomic DNA is analyzed for *FOXC2* mutations by DNA sequencing of the coding exons of the *FOXC2* 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**

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<thead>
<tr>
<th></th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<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 exon of *FOXC2*.

**SPECIMEN REQUIREMENTS**

- **Blood (preferred):** EDTA (purple-top) tubes: *Adult:* 5 cc *Child:* 5 cc *Infant:* 2-3 cc
- **Tissue:** Frozen (preferred), RNA*later*
- **Other Body Fluids or Formalin-fixed, Paraffin-embedded Tissue:** Call to inquire

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