GATA6 (GATA Binding Protein 6) is a member of a small family of zinc finger transcription factors that play an important role in the regulation of cellular differentiation and organogenesis during vertebrate development. GATA6 gene contains 6 exons and spans 33.086 Kb genome sequence on chromosome 18q11-q12, and is expressed during early embryogenesis and localizes to endo- and mesodermally derived cells during later embryogenesis and thereby plays an important role in gut, lung, and heart development. Mutations in this gene are associated with several congenital defects including atrioventricular septal defect 5 and atrial septal defect 9. GATA6 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 GATA6 mutations. Individuals are tested by DNA sequencing of the coding exons of the GATA6 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 GATA6 mutations by DNA sequencing of the coding exons of the GATA6 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></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>$600 per sample</td>
<td>81405</td>
</tr>
<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 2-7 of GATA6.

**SPECIMEN REQUIREMENTS**

<table>
<thead>
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<th>Blood (preferred): EDTA (purple-top) tubes</th>
<th>Adult: 5 cc</th>
<th>Child: 5 cc</th>
<th>Infant: 2-3 cc</th>
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<tr>
<td>Tissue: Frozen (preferred), RNAlater</td>
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<tr>
<td>Other Body Fluids or Formalin-fixed, Paraffin-embedded Tissue</td>
<td>Call to inquire</td>
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</tbody>
</table>

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