The proliferation and differentiation of B cells is dependent upon a B-cell antigen receptor (BCR) complex. Binding of antigens to specific B-cell receptors results in a tyrosine phosphorylation reaction through the BCR complex and leads to multiple signal transduction pathways. *IGBP1* (immunoglobulin binding protein 1) gene encodes the protein which is part of surface IgM-receptor. *IGBP1* gene contains 6 coding exons covering around 32.87 Kb genomic sequences and has been mapped to chromosome Xq13.1-q13.3. Mutations in *IGBP1* gene are linked to agenesis of the corpus callosum, with mental retardation, ocular coloboma and micrognathia and agenesis of the corpus callosum - intellectual disability - coloboma - micrognathia. *IGBP1* mutations demonstrate X-linked recessive 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 *IGBP1* mutations. Individuals are tested by DNA sequencing of the coding exons of the *IGBP1* 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.

**METHODOLOGY**

Genomic DNA is analyzed for *IGBP1* mutations by DNA sequencing of the coding exons of the *IGBP1* 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><strong>Direct and Institutional Billing</strong></th>
<th><strong>CPT Codes</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Index Case (Male or Female)</strong></td>
<td>$600 per sample</td>
<td>81405</td>
</tr>
<tr>
<td><strong>Additional Family Members</strong></td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
</tr>
</tbody>
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**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 2-7 of *IGBP1*.

**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 or Formalin-fixed, Paraffin-embedded Tissue: Call to inquire