**TBX20 MUTATION ANALYSIS**

**JOHN WELSH CARDIOVASCULAR DIAGNOSTIC LABORATORY**

TBX20, like TBX5, is a member of the phylogenetically conserved family of T-box genes. T-box is a common DNA-binding domain. Human TBX20 encodes a deduced 297-amino acid protein that shares approximately 71% sequence identity with the T-box domain of the Drosophila H15 protein. RT-PCR analysis showed that human TBX20 is expressed in the fetal heart, eye and limb, indicating a possible role in regulating development of these tissues. TBX20 gene contains eight exons spanning 22 kb of genomic distance that was mapped to chromosome 7p15-p14. Multiple mutations in TBX20 have been identified in patients with atrial septal defect 4 (ASD4), ventricular septal defect (VSD), patent foramen ovale and cardiac valve defects and dilated cardiomyopathy. TBX20 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 TBX20 mutations. Individuals are tested by DNA sequencing of the coding exons of the TBX20 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 ASD, VSD, patent foramen ovale and cardiac valve defects

**METHODOLOGY**

Genomic DNA is analyzed for TBX20 mutations by DNA sequencing of the coding exons of the TBX20 gene, as well as the exon/intron junctions and a portion of the 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>Service</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>$750 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% detection of mutations in the coding exons of TBX20

**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