BCOR gene encodes BCL6 corepressor (BCOR), a POZ/zinc finger transcription repressor that is required for germinal center formation and may influence apoptosis. Research has shown that mutations in this gene result in microphthalmia, syndromic 2 (MCOPS2) and oculofaciocardiodental syndrome (OFCD). Mapped to chromosome Xp11.4, BCOR gene spans a genomic distance of approximately 127 kb, is composed of 15 exons, and has twelve splice variants. BCOR mutations demonstrate X-linked inheritance; however, definitive genotype/phenotype correlations have not yet been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for BCOR mutations. Individuals are tested by DNA sequencing of the coding exons of the BCOR 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 microphthalmia, syndromic 2 (MCOPS2) or oculofaciocardiodental syndrome (OFCD).

METHODOLOGY

Genomic DNA is analyzed for BCOR mutations by DNA sequencing of the coding exons of the BCOR 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>
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<tr>
<th>Service</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>$1,150 per sample;</td>
<td>81406</td>
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<tr>
<td>Additional Family Members</td>
<td>$300 per sample;</td>
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<td>known familial mutation only</td>
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SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 2-15 of BCOR.

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