B3GAT3 MUTATION ANALYSIS
JOHN WELSH CARDIOVASCULAR
DIAGNOSTIC LABORATORY

B3GAT3 gene encodes beta-1,3-glucuronyltransferase 3 (glucuronosyltransferase 1), an enzyme that exhibits strict acceptor specificity, recognizing non-reducing terminal sugars and their anomeric linkages. Research has shown that mutations in this gene result in multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, and mucopolysaccharidoses. Mapped to chromosome 11q12.3, B3GAT3 gene spans a genomic distance of approximately 6.8 kb, is composed of 5 exons, and has six splice variants. B3GAT3 mutations demonstrate autosomal recessive inheritance; however, definitive genotype/phenotype correlations have not yet been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for B3GAT3 mutations. Individuals are tested by DNA sequencing of the coding exons of the B3GAT3 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 multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, or mucopolysaccharidoses.

METHODOLOGY
Genomic DNA is analyzed for B3GAT3 mutations by DNA sequencing of the coding exons of the B3GAT3 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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<tr>
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<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>
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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 exons 1-5 of B3GAT3.

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 and Formalin-fixed, Paraffin-embedded Tissue: Call to inquire