CHD7 MUTATION ANALYSIS
JOHN WELSH CARDIOVASCULAR
DIAGNOSTIC LABORATORY

CHD7 gene encodes chromodomain helicase DNA-binding protein 7 (CHD7), a protein that contains several helicase family domains. Research has shown that mutations in this gene result in CHARGE syndrome, hypogonadotropic hypogonadism 5 with or without anosmia, and scoliosis, idiopathic 3. Mapped to chromosome 8q12.1-q12.2, CHD7 gene spans a genomic distance of approximately 189 kb, is composed of 38 exons, and has twelve splice variants. CHD7 mutations demonstrate autosomal dominant inheritance; however, definitive genotype/phenotype correlations have not yet been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for CHD7 mutations. Individuals are tested by DNA sequencing of the coding exons of the CHD7 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, CHARGE syndrome, hypogonadotropic hypogonadism 5 with or without anosmia, or scoliosis, idiopathic 3.

METHODOLOGY

Genomic DNA is analyzed for CHD7 mutations by DNA sequencing of the coding exons of the CHD7 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>$1,350 per sample</td>
<td>81407</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 2-38 of CHD7.

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

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