ADCK3 MUTATION ANALYSIS
JOHN WELSH CARDIOVASCULAR
DIAGNOSTIC LABORATORY

ADCK3 gene encodes for a mitochondrial protein similar to yeast ABC1, which functions in an electron-transferring membrane protein complex in the respiratory chain. Research has shown that mutations in this gene result in coenzyme Q10 deficiency, primary, 4 and spinocerebellar ataxia, autosomal recessive, 9. Mapped to chromosome 1q42.13, ADCK3 gene spans a genomic distance of approximately 90 kb, is composed of 15 exons, and has nine splice variants. ADCK3 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 ADCK3 mutations. Individuals are tested by DNA sequencing of the coding exons of the ADCK3 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, primary coenzyme Q10 deficiency 4 and autosomal recessive spinocerebellar ataxia 9.

METHODOLOGY
Genomic DNA is analyzed for ADCK3 mutations by DNA sequencing of the coding exons of the ADCK3 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>$1,100 per sample</td>
<td>81406</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 2-15 of ADCK3.

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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