The DOLK gene is located at 9q34.11 and encodes 538 amino acid dolichyl monophosphate, which is an essential glycosyl carrier lipid for C- and O-mannosylation and N-glycosylation of proteins and for biosynthesis of glycosyl phosphatidylinositol anchors in endoplasmic reticulum (ER). Dolk catalyzes the CTP-mediated phosphorylation of dolichol, the terminal step in de novo dolichyl monophosphate biosynthesis DOLK mutations are passed on in an autosomal recessive fashion. Mutations in this gene are associated with congenital disorder of glycosylation, type Im, and cardiomyopathy. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for DOLK mutations. Individuals are tested by DNA sequencing of the coding exons of the DOLK 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 congenital disorder of glycosylation, type Im, and dilated cardiomyopathy.

METHODOLOGY

Genomic DNA is analyzed for DOLK mutations by DNA sequencing of the coding exons of the DOLK 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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<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<td>Index Case (Male or Female)</td>
<td>$300 per sample</td>
<td>81403</td>
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<tr>
<td>Additional Family Members</td>
<td>$300 per sample; Known familial mutation only</td>
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SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exon 1 of DOLK.

SPECIMEN REQUIREMENTS

Blood (preferred): EDTA (purple-top) tubes: Adult: 5 cc  Child: 5 cc  Infant: 2-3 cc
Tissue: Frozen (preferred) or RNA later
Other Body Fluids or Formalin-Fixed, Paraffin-Embedded Tissue: Call to inquire