LDB3 – Associated Dilated Cardiomyopathy

LDB3 – Associated Dilated Cardiomyopathy is characterized by left ventricular enlargement and systolic dysfunction. While there are more than 20 genes associated with autosomal dominant dilated cardiomyopathy, LDB3 mutations are thought to comprise a very small number. The LDB3 gene codes for the LIM domain binding protein-3 which is a cytoskeletal protein. The LDB3 gene contains 16 exons and is located on chromosome 10q22.2-q23.3.

In addition to causing autosomal dominant dilated cardiomyopathy, LDB3 is unique in that it can also be associated with left ventricular noncompaction and myopathy (1).

Indication

LDB3 gene testing is utilized to confirm a diagnosis of dilated cardiomyopathy in patients with clinically evident disease. Genetic testing allows for early identification and diagnosis of individuals at greatest risk prior to the expression of typical clinical manifestations.
Methodology:

All 16 exons of the LDB3 gene, as well as the exon/intron boundaries and portion of untranslated regions of the gene are amplified by PCR. Genomic DNA sequences from both forward and reverse directions are obtained by automatic fluorescent detection using an ABI PRISM® 3730 DNA Analyzer. Sequence variants different from National Center for Biotechnology Information GenBank references are further evaluated for genetic significance. If a mutation is identified, a known familial mutation analysis will be available for additional family members.

Sensitivity & Accuracy:

Greater than 98.5% of the mutations in exons 1-16 of LDB3 are detectable by sequence based methods. Sequencing does not detect deletions or duplications.

References:


Specimen:

Peripheral blood in EDTA tube
Adult: 5-10mL
Child: 3-5mL
Infant: 1-3mL
For other specimen types, please contact Amy Shikany at 513-803-3317

Turnaround Time:

Full Mutation Analysis 2-4 weeks
Known Mutation Analysis 1-2 weeks

CPT Codes:

Full Gene Sequencing 81406
Additional Family Members 81403