Cincinnati Children's
Heart Institute

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[1]. 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.

Heart Institute Diagnostic Lab
CAP#: 7518730
CLIA#: 36D2003208
Phone: (513) 803-1751
Fax: (513) 803-1748
Email: HeartDx@cchmc.org

Shipping Instructions

Please enclose a test requisition form with sample. All information must be complete before sample can be processed. Samples may be shipped at room temperature by overnight Federal Express to arrive Monday through Friday.

Ship To:

Cincinnati Children's
Hospital Medical Center
Attn: Heart Institute Diagnostic Lab
240 Albert Sabin Way,
Room S4.381
Cincinnati, OH 45229-3039

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:

1. Vatta M, Mohapatra B, Jimenez S, Sanchez X, Faulkner G, Perles Z, Sinagra G, Lin JH, Vu TM, Zhou Q, Bowles KR, Di Lenarda A, Schimmenti L, Fox M, Chrisco MA, Murphy RT, McKenna W, Elliott P, Bowles NE, Chen J, Valle G, Towbin JA. Mutations in Cypher/ZASP in patients with dilated cardiomyopathy and left ventricular non-compaction. *Journal of the American College of Cardiology*. 2003;42:2014-2027.

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