LDB3 gene

LIM domain binding 3

Normal Function

The *LDB3* gene provides instructions for making a protein that is found in heart (cardiac) muscle and in the muscles used for movement (skeletal muscle). Within muscle fibers, structures called sarcomeres generate the force needed for muscles to contract. Sarcomeres are linked together by structures called Z-discs to form myofibrils, which are the basic units of muscle fibers. The LDB3 protein helps to stabilize the sarcomeres by interacting with Z-discs. This organization of the sarcomeres and myofibrils provides the strength and stability that is needed during repeated cycles of muscle contraction and relaxation.

Health Conditions Related to Genetic Changes

Myofibrillar myopathy

Variants (also called mutations) in the *LDB3* gene have been found to cause myofibrillar myopathy. This condition is characterized by muscle weakness (myopathy) that worsens over time. Many of the *LDB3* variants that cause myofibrillar myopathy lead to the substitution of one protein building block (amino acid) for another in the LDB3 protein. The altered protein cannot properly interact with Z-discs, which impairs the organization and assembly of myofibrils. Myofibrils that are not organized correctly break down and form clumps (aggregates) of abnormal proteins within the sarcomere, which leads to the myopathy seen in people with myofibrillar myopathy.

Nonsyndromic dilated cardiomyopathy

MedlinePlus Genetics provides information about Nonsyndromic dilated cardiomyopathy

Familial hypertrophic cardiomyopathy

MedlinePlus Genetics provides information about Familial hypertrophic cardiomyopathy

Left ventricular noncompaction

MedlinePlus Genetics provides information about Left ventricular noncompaction

Other Names for This Gene

- cypher
- KIAA0613
- LIM domain-binding protein 3
- oracle
- PDLIM6
- Z-band alternatively spliced PDZ motif protein
- ZASP

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of LDB3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=11155[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28LDB3%5BTIAB%5D%29+OR+%28LIM+domain+binding+3%5BTIAB%5D%29%29+OR+%28ZASP%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- LIM DOMAIN-BINDING 3; LDB3 (https://omim.org/entry/605906)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/11155)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=LDB3[gene])

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Genomic Location

The *LDB3* gene is found on chromosome 10 (https://medlineplus.gov/genetics/chromosome/10/).

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