



## LDHA gene

lactate dehydrogenase A

### Normal Function

The *LDHA* gene provides instructions for making a protein called lactate dehydrogenase-A, which is one piece (subunit) of the lactate dehydrogenase enzyme. There are five different forms of this enzyme, each made up of four protein subunits. Various combinations of lactate dehydrogenase-A subunits and lactate dehydrogenase-B subunits (which are produced from a different gene) make up the different forms of the enzyme. The lactate dehydrogenase enzyme is found throughout the body and is important for performing a chemical reaction that provides energy for the body.

The version of lactate dehydrogenase made of four lactate dehydrogenase-A subunits is found primarily in skeletal muscles, which are muscles used for movement. Skeletal muscles need large amounts of energy during high-intensity physical activity when the body's oxygen intake is not sufficient for the amount of energy required (anaerobic exercise). To create additional energy, glucose stored in the body as glycogen gets broken down. During the final stage of glycogen breakdown, the lactate dehydrogenase enzyme converts the molecule pyruvate into a similar molecule called lactate, which can be used by the body for energy.

### Health Conditions Related to Genetic Changes

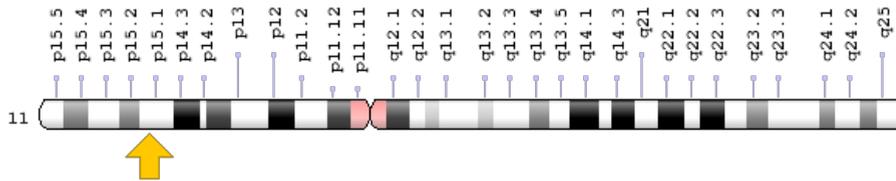
#### Lactate dehydrogenase deficiency

At least eight mutations in the *LDHA* gene have been found to cause lactate dehydrogenase deficiency, specifically lactate dehydrogenase-A deficiency. This condition is characterized by extreme tiredness and the breakdown of muscle tissue following high-intensity exercise or other strenuous activity. *LDHA* gene mutations result in the production of an abnormal lactate dehydrogenase-A subunit that cannot attach (bind) to other subunits to form lactate dehydrogenase enzyme. A lack of functional subunit reduces the amount of enzyme that is formed, mostly affecting skeletal muscles. As a result, glycogen is not broken down efficiently, leading to decreased energy in muscle cells. When muscle cells do not get sufficient energy during strenuous physical activity, the muscles become weak and muscle tissue can break down, as experienced by people with lactate dehydrogenase-A deficiency.

## Chromosomal Location

Cytogenetic Location: 11p15.1, which is the short (p) arm of chromosome 11 at position 15.1

Molecular Location: base pairs 18,394,389 to 18,408,425 on chromosome 11 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- L-lactate dehydrogenase A chain
- lactate dehydrogenase M
- LDH-A
- LDH-M
- LDH muscle subunit
- LDH1
- LDHA\_HUMAN
- LDHM

## Additional Information & Resources

### Educational Resources

- Biochemistry (fifth edition, 2002): Isozymes of Lactate Dehydrogenase  
<https://www.ncbi.nlm.nih.gov/books/NBK22392/figure/A1362/>
- Biochemistry (fifth edition, 2002): Lactate and Alanine Formed by Contracting Muscle Are Used by Other Organs  
<https://www.ncbi.nlm.nih.gov/books/NBK22423/#A2286>

- Research Collaboratory for Structural Bioinformatics Protein Data Bank: Lactate Dehydrogenase  
<http://pdb101.rcsb.org/motm/102>
- Washington University, St. Louis: Neuromuscular Disease Center: Lactate Dehydrogenase A Deficiency  
<https://neuromuscular.wustl.edu/msys/glycogen.html#ldh>

#### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28LDHA%5BTI%5D%29+OR+%28lactate+dehydrogenase+A%5BTI%5D%29+OR+%28lactate+dehydrogenase+M%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

#### Catalog of Genes and Diseases from OMIM

- LACTATE DEHYDROGENASE A  
<http://omim.org/entry/150000>

#### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_LDHA.html](http://atlasgeneticsoncology.org/Genes/GC_LDHA.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=LDHA%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:6535](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6535)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:3939>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/3939>
- UniProt  
<https://www.uniprot.org/uniprot/P00338>

#### **Sources for This Summary**

- Kanno T, Maekawa M. Lactate dehydrogenase M-subunit deficiencies: clinical features, metabolic background, and genetic heterogeneities. *Muscle Nerve Suppl.* 1995;3:S54-60.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/7603529>
- OMIM: LACTATE DEHYDROGENASE A  
<http://omim.org/entry/150000>

- Lee BJ, Zand L, Manek NJ, Hsiao LL, Babovic-Vuksanovic D, Wylam ME, Qian Q. Physical therapy-induced rhabdomyolysis and acute kidney injury associated with reduced activity of muscle lactate dehydrogenase A. *Arthritis Care Res (Hoboken)*. 2011 Dec;63(12):1782-6. doi: 10.1002/acr.20584. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/22127970>
  - Miyajima H, Takahashi Y, Kaneko E. Characterization of the glycolysis in lactate dehydrogenase-A deficiency. *Muscle Nerve*. 1995 Aug;18(8):874-8. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/7630349>
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<https://ghr.nlm.nih.gov/gene/LDHA>

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