LAMA2 gene
laminin subunit alpha 2

Normal Function

The LAMA2 gene provides instructions for making a part (subunit) of certain members of a protein family called laminins. Laminin proteins are made of three different subunits called alpha, beta, and gamma. There are several forms of each subunit, and each form is produced from instructions carried by a different gene. The LAMA2 gene provides instructions for the alpha-2 subunit. This subunit, together with the beta-1 and gamma-1 subunits, forms the laminin 2 protein, also known as merosin or laminin-211. The alpha-2 subunit, along with the beta-2 and gamma-1 subunits, also forms another laminin called laminin 4, sometimes known as laminin-221.

Laminins are found in an intricate lattice of proteins and other molecules that forms in the spaces between cells (the extracellular matrix). There, the laminins help regulate cell growth, cell movement (motility), and the attachment of cells to one another (adhesion). They are also involved in the formation and organization of basement membranes, which are thin, sheet-like structures within the extracellular matrix that separate and support cells in many tissues. Laminin 2 and laminin 4 play a particularly important role in the muscles used for movement (skeletal muscles). The laminins attach (bind) to other proteins in the extracellular matrix and in the membrane of muscle cells, which helps maintain the stability of muscle fibers.

Health Conditions Related to Genetic Changes

LAMA2-related muscular dystrophy

More than 100 LAMA2 gene mutations have been identified in individuals with LAMA2-related muscular dystrophy, a disorder that causes weakness and wasting (atrophy) of skeletal muscles. This condition generally appears in one of two ways: as a severe, early-onset type or a milder, late-onset form. Most LAMA2 gene mutations that cause early-onset LAMA2-related muscular dystrophy result in the absence of functional laminin alpha-2 subunit. Mutations that cause late-onset LAMA2-related muscular dystrophy usually result in a reduction (deficiency) of functional laminin alpha-2 subunit. Deficiency or absence of the laminin alpha-2 subunit results in a corresponding lack of laminin 2 and laminin 4, reducing the strength and stability of muscle tissue and leading to the signs and symptoms of LAMA2-related muscular dystrophy.

Limb-girdle muscular dystrophy
Chromosomal Location

Cytogenetic Location: 6q22.33, which is the long (q) arm of chromosome 6 at position 22.33

Molecular Location: base pairs 128,883,138 to 129,516,566 on chromosome 6 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- LAMA2_HUMAN
- laminin-2 subunit alpha
- laminin-4 subunit alpha
- laminin-12 subunit alpha
- laminin M chain
- laminin subunit alpha-2
- laminin subunit alpha-2 isoform a precursor
- laminin subunit alpha-2 isoform b precursor
- laminin, alpha 2
- LAMM
- merosin heavy chain

Additional Information & Resources

Clinical Information from GeneReviews

- LAMA2-Related Muscular Dystrophy
  https://www.ncbi.nlm.nih.gov/books/NBK97333
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28LAMA2%5BTIAB%5D%29+OR+%28%28laminin+M+chain%5BTIAB%5D%29+OR+%28merosin+heavy+chain+5BTIAB%5D%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

- LAMININ, ALPHA-2
  http://omim.org/entry/156225

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_LAMA2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=LAMA2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6482
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3908
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P24043

Sources for This Summary

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11584042
Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1734735/

• OMIM: LAMININ, ALPHA-2
http://omim.org/entry/156225

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10679965

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