



LAMC2 gene

laminin subunit gamma 2

Normal Function

The *LAMC2* gene provides instructions for making one part (subunit) of a protein called laminin 332 (formerly known as laminin 5). This protein is made up of three subunits, called alpha, beta, and gamma. The *LAMC2* gene carries instructions for the gamma subunit; the alpha and beta subunits are produced from other genes.

Laminins are a group of proteins that 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 that separate and support cells in many tissues. Laminin 332 has a particularly important role in the basement membrane that underlies the top layer of skin (the epidermis). This membrane gives strength and resiliency to the skin and creates an additional barrier between the body and its surrounding environment. Laminin 332 is a major component of fibers called anchoring filaments, which connect the two layers of the basement membrane and help hold the skin together.

Studies suggest that laminin 332 also has several other functions. This protein appears to be important for wound healing. Additionally, researchers have proposed roles for laminin 332 in the clear outer covering of the eye (the cornea) and in the development of tooth enamel.

Health Conditions Related to Genetic Changes

Junctional epidermolysis bullosa

More than 40 mutations in the *LAMC2* gene have been identified in people with junctional epidermolysis bullosa (JEB). The more serious form of the disease, known as JEB generalized severe, usually results from mutations that prevent the production of functional laminin 332. Most of these mutations lead to a premature stop signal in the instructions for making the gamma subunit of laminin 332, which prevents the assembly of this protein. Without laminin 332, the epidermis is only weakly connected to the underlying layers of skin. Friction or other minor trauma (such as rubbing or scratching) can cause the skin layers to separate, leading to the formation of blisters. Infants with JEB generalized severe develop widespread blistering that causes life-threatening complications.

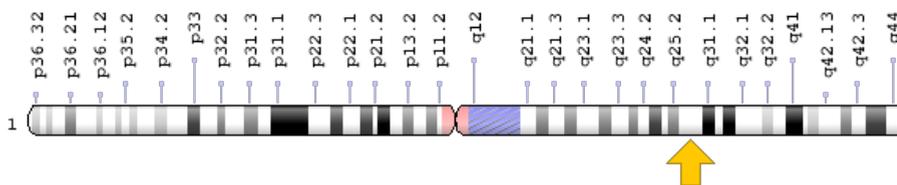
Other *LAMC2* gene mutations cause the milder form of junctional epidermolysis bullosa, JEB generalized intermediate. Some of these mutations alter single protein building blocks (amino acids) in the gamma subunit of laminin 332. Others add or remove a small number of amino acids in the gamma subunit or change the way the

gene's instructions are used to make the subunit. The genetic changes responsible for JEB generalized intermediate usually lead to the production of a laminin 332 protein that retains some of its function. Affected individuals experience blistering, but it may be limited to the hands, feet, knees, and elbows and often improves after the newborn period.

Chromosomal Location

Cytogenetic Location: 1q25.3, which is the long (q) arm of chromosome 1 at position 25.3

Molecular Location: base pairs 183,186,264 to 183,245,127 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- B2T
- BM600
- BM600-100kDa
- cell-scattering factor (140kDa)
- CSF
- EBR2
- EBR2A
- kalinin (105kD)
- kalinin-105kDa
- ladsin (140kDa)
- LAM5, gamma-2 subunit
- LAMB2T
- LAMC2_HUMAN
- laminin 5, gamma-2 subunit

- laminin, gamma 2
- laminin, gamma-2
- laminin, nicein, beta-2
- LAMNB2
- MGC138491
- MGC141938
- nicein (100kDa)
- nicein-100kDa

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Structure of Laminin (image)
<https://www.ncbi.nlm.nih.gov/books/NBK21706/?rendertype=figure&id=A6568>

Clinical Information from GeneReviews

- Junctional Epidermolysis Bullosa
<https://www.ncbi.nlm.nih.gov/books/NBK1125>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28LAMC2%5BTIAB%5D%29+OR+%28laminin+%5Btiab%5D+AND+gamma+2+%5Btiab%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- LAMININ, GAMMA-2
<http://omim.org/entry/150292>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_LAMC2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=LAMC2%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6493
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:3918>

- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3918>
- UniProt
<https://www.uniprot.org/uniprot/Q13753>

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Reviewed: July 2019

Published: November 12, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services