



## ADAMTSL2 gene

ADAMTS like 2

### Normal Function

The *ADAMTSL2* gene provides instructions for making a protein whose function is unknown. The ADAMTSL2 protein is active in many different tissues. It is found in the extracellular matrix, which is the intricate lattice of proteins and other molecules that forms in the spaces between cells.

Studies suggest that the ADAMTSL2 protein interacts with a protein called latent transforming growth factor beta binding protein 1 (LTBP1). The LTBP1 protein is involved in the storage of transforming growth factor beta (TGF- $\beta$ ), a critical growth factor that helps control the growth and division (proliferation) of cells, the process by which cells mature to carry out specific functions (differentiation), cell movement (motility), and the self-destruction of cells (apoptosis). Through its interaction with the LTBP1 protein, researchers suspect that the ADAMTSL2 protein may help regulate the availability of TGF- $\beta$ .

The interaction between the ADAMTSL2 protein and the LTBP1 protein suggests that ADAMTSL2 may also play a role in the microfibrillar network. This organized clustering of thread-like filaments (called microfibrils) in the extracellular matrix provides strength and flexibility to tissues throughout the body.

### Health Conditions Related to Genetic Changes

#### geleophysic dysplasia

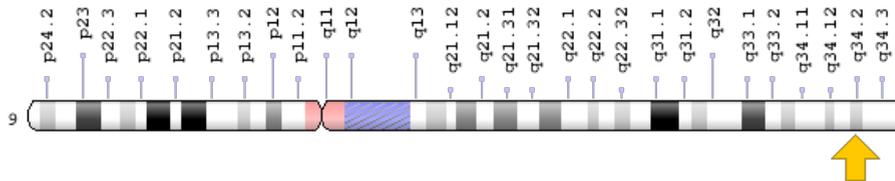
At least five mutations in the *ADAMTSL2* gene have been identified in people with geleophysic dysplasia. Most of these mutations change single protein building blocks (amino acids) in the ADAMTSL2 protein, which probably alters the protein's 3-dimensional shape.

Through a process that is poorly understood, *ADAMTSL2* gene mutations alter the microfibrillar network in many different tissues. Impairment of this essential network disrupts the normal functions of cells, which likely contributes to the varied signs and symptoms of geleophysic dysplasia. Researchers are working to determine how mutations in the *ADAMTSL2* gene lead to short stature, heart disease, and the other features of this condition.

## Chromosomal Location

Cytogenetic Location: 9q34.2, which is the long (q) arm of chromosome 9 at position 34.2

Molecular Location: base pairs 133,532,164 to 133,575,519 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ADAMTS-like 2
- ADAMTS-like 2 precursor
- ATL2\_HUMAN
- FLJ45164
- KIAA0605

## Additional Information & Resources

### Educational Resources

- Developmental Biology (sixth edition, 2000): The TGF- $\beta$  Superfamily  
<https://www.ncbi.nlm.nih.gov/books/NBK10071/#A1046>

### GeneReviews

- Geleophysic Dysplasia  
<https://www.ncbi.nlm.nih.gov/books/NBK11168>

### Genetic Testing Registry

- GTR: Genetic tests for ADAMTSL2  
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9719%5Bgeneid%5D>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ADAMTSL2%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+3600+days%22%5Bdp%5D>

## OMIM

- ADAMTS-LIKE PROTEIN 2  
<http://omim.org/entry/612277>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ADAMTSL2.html](http://atlasgeneticsoncology.org/Genes/GC_ADAMTSL2.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ADAMTSL2%5Bgene%5D>
- HGNC Gene Family: ADAMTS like  
<http://www.genenames.org/cgi-bin/genefamilies/set/947>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=14631](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=14631)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/9719>
- UniProt  
<http://www.uniprot.org/uniprot/Q86TH1>

## **Sources for This Summary**

- GeneReview: Geleophysic Dysplasia  
<https://www.ncbi.nlm.nih.gov/books/NBK11168>
- Koo BH, Le Goff C, Jungers KA, VasANJI A, O'Flaherty J, Weyman CM, Apte SS. ADAMTS-like 2 (ADAMTSL2) is a secreted glycoprotein that is widely expressed during mouse embryogenesis and is regulated during skeletal myogenesis. *Matrix Biol.* 2007 Jul;26(6):431-41. Epub 2007 Mar 30.  
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- Le Goff C, Morice-Picard F, Dagoneau N, Wang LW, Perrot C, Crow YJ, Bauer F, Flori E, Prost-Squarcioni C, Krakow D, Ge G, Greenspan DS, Bonnet D, Le Merrer M, Munnich A, Apte SS, Cormier-Daire V. ADAMTSL2 mutations in geleophysic dysplasia demonstrate a role for ADAMTS-like proteins in TGF-beta bioavailability regulation. *Nat Genet.* 2008 Sep;40(9):1119-23. doi: 10.1038/ng.199.  
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