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-β), 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-β.

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 Updated Annotation Release 109.20190607, GRCh38.p13) (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-β Superfamily
  https://www.ncbi.nlm.nih.gov/books/NBK10071/#A1046

**Clinical Information from GeneReviews**

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

**Scientific Articles on PubMed**

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ADAMTSL2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22+AND+human%5Bmh%5D
Catalog of Genes and Diseases from 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

• ClinVar

• HGNC Gene Symbol Report

• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:9719

• NCBI Gene

• UniProt
  https://www.uniprot.org/uniprot/Q86TH1

Sources for This Summary


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