

## 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.

## Other Names for This Gene

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

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of ADAMTSL2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9719\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9719[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29>)

### Catalog of Genes and Diseases from OMIM

- ADAMTS-LIKE PROTEIN 2; ADAMTSL2 (<https://omim.org/entry/612277>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/9719>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ADAMTSL2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ADAMTSL2[gene]))

## References

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## Genomic Location

The *ADAMTSL2* gene is found on chromosome 9 (<https://medlineplus.gov/genetics/chromosome/9/>).

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