

ADAMTS2 gene

ADAM metalloproteinase with thrombospondin type 1 motif 2

Normal Function

The *ADAMTS2* gene provides instructions for making an enzyme that processes several types of procollagen molecules. Procollagens are the precursors of collagens, which are complex molecules found in the spaces between cells that add strength, support, and stretchiness (elasticity) to many body tissues. The ADAMTS2 enzyme cuts a short chain of protein building blocks (amino acids) off one end of procollagens. This clipping step is necessary for the resulting collagen molecules to assemble into strong, slender fibrils.

Health Conditions Related to Genetic Changes

Ehlers-Danlos syndrome

Several mutations in the *ADAMTS2* gene have been identified in people with a form of Ehlers-Danlos syndrome called the dermatosparaxis type. Ehlers-Danlos syndrome is a group of disorders that affect the connective tissues supporting the skin, bones, blood vessels, and many other organs and tissues. The dermatosparaxis type is characterized by soft, fragile skin that sags and wrinkles; easy bruising; and distinctive facial features.

Mutations in the *ADAMTS2* gene greatly reduce the production or activity of the ADAMTS2 enzyme. Without enough of this enzyme, procollagens cannot be processed correctly. As a result, collagen fibrils are not assembled properly. The resulting fibrils are disorganized, which weakens connective tissues and leads to the signs and symptoms of the disorder.

Other Names for This Gene

- a disintegrin and metalloproteinase with thrombospondin motifs 2
- a disintegrin-like and metalloprotease (reprolysin type) with thrombospondin type 1 motif, 2
- ADAM metalloproteinase with thrombospondin type 1 motif, 2
- ADAM-TS2
- ATS2_HUMAN
- hPCPNI

- NPI
- PCINP
- PCPNI
- pNPI
- procollagen I N-proteinase
- procollagen I/II amino-propeptide processing enzyme
- procollagen N-endopeptidase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ADAMTS2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9509\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9509[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ADAMTS2%5BTIAB%5D%29+OR+%28%28procollagen+I+N-proteinase%5BTIAB%5D%29+OR+%28Procollagen+N-endopeptidase%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+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- A DISINTEGRIN-LIKE AND METALLOPROTEINASE WITH THROMBOSPONDIN TYPE 1 MOTIF, 2; ADAMTS2 (<https://omim.org/entry/604539>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/9509>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ADAMTS2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ADAMTS2[gene]))

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Genomic Location

The *ADAMTS2* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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