ADAMTS2 gene

ADAM metallopeptidase 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

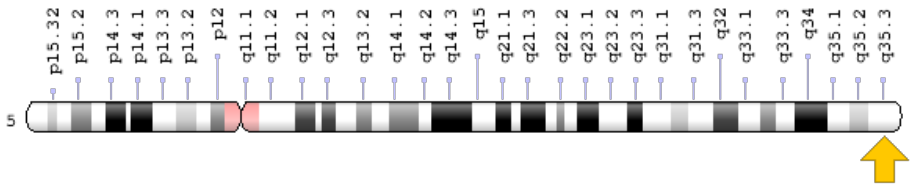
Several mutations in the *ADAMTS2* gene have been identified in people with a form of Ehlers-Danlos syndrome called the dermatosparaxis type. Ehlers-Danlos syndrome is a group of disorders that affect the connective tissues that support the skin, bones, blood vessels, and many other organs and tissues. 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.

Chromosomal Location

Cytogenetic Location: 5q35.3, which is the long (q) arm of chromosome 5 at position 35.3

Molecular Location: base pairs 179,110,851 to 179,345,430 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

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 metallopeptidase 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

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): After Secretion, Fibrillar Procollagen Molecules Are Cleaved to Collagen Molecules, Which Assemble into Fibrils
  https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3557
- Molecular Cell Biology (fourth edition, 2000): Collagen: The Fibrous Proteins of the Matrix
  https://www.ncbi.nlm.nih.gov/books/NBK21582/

Genetic Testing Registry

- GTR: Genetic tests for ADAMTS2
  https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9509%5Bgeneid%5D

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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

OMIM

- A DISINTEGRIN-LIKE AND METALLOPROTEINASE WITH THROMBOSPONDIN TYPE 1 MOTIF, 2
  http://omim.org/entry/604539

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ADAMTS2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ADAMTS2%5Bgene%5D
- Ehlers-Danlos Syndrome Variant Database
  https://eds.gene.le.ac.uk/home.php?select_db=ADAMTS2
- HGNC Gene Family: ADAM metallopeptidases with thrombospondin type 1 motif
  http://www.genenames.org/cgi-bin/genefamilies/set/50

- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=218
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/9509
- UniProt
  http://www.uniprot.org/uniprot/O95450

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