ADAMTS10 gene

ADAM metallopeptidase with thrombospondin type 1 motif 10

Normal Function

The *ADAMTS10* gene provides instructions for making an enzyme that is found in many of the body's cells and tissues. This enzyme is part of a family of metalloproteases, which are zinc-containing enzymes that cut apart other proteins. Although the function of the ADAMTS10 enzyme is unknown, it is critical for growth before and after birth. Researchers believe that it may be involved in the development of structures including the skin, eyes, heart, and skeleton.

Health Conditions Related to Genetic Changes

Weill-Marchesani syndrome

At least five mutations in the *ADAMTS10* gene have been identified in people with Weill-Marchesani syndrome. Each of these mutations prevents the cell from producing any functional ADAMTS10 enzyme. Researchers speculate that a loss of this enzyme disrupts skeletal development, leading to short stature and unusually short fingers and toes (brachydactyly). A shortage of the ADAMTS10 enzyme also interferes with the development and function of the lens of the eye, causing eye abnormalities and impaired vision. Additionally, a lack of this enzyme may disrupt the normal development of the heart, resulting in the heart defects occasionally seen in people with Weill-Marchesani syndrome.

Other Names for This Gene

- a disintegrin and metalloproteinase with thrombospondin motifs 10
- a disintegrin-like and metalloprotease (reprolysin type) with thrombospondin type 1 motif, 10
- a disintegrin-like and metalloprotease domain with thrombospondin type I repeats 10
- ADAM metallopeptidase with thrombospondin type 1 motif, 10
- ADAM-TS10
- ADAMTS-10
- ATS10_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ADAMTS10 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=81794[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ADAMTS10%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- A DISINTEGRIN-LIKE AND METALLOPROTEINASE WITH THROMBOSPONDIN TYPE 1 MOTIF, 10; ADAMTS10 (https://omim.org/entry/608990)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/81794)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ADAMTS10[gene])

References

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- Kutz WE, Wang LW, Dagoneau N, Odrcic KJ, Cormier-Daire V, Traboulsi EI, ApteSS. Functional analysis of an ADAMTS10 signal peptide mutation inWeill-Marchesani syndrome demonstrates a long-range effect on secretion of thefull-length enzyme. Hum Mutat. 2008 Dec;29(12):1425-34. doi: 10.1002/humu.20797. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18567016)
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- Somerville RP, Jungers KA, Apte SS. Discovery and characterization of a novel, widely expressed metalloprotease, ADAMTS10, and its proteolytic activation. JBiol Chem. 2004 Dec 3;279(49):51208-17. doi: 10.1074/jbc.M409036200. Epub 2004Sep

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Genomic Location

The *ADAMTS10* gene is found on chromosome 19 (https://medlineplus.gov/genetics/chromosome/19/).

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