



ADAMTS13 gene

ADAM metalloproteinase with thrombospondin type 1 motif 13

Normal Function

The *ADAMTS13* gene provides instructions for making an enzyme that is involved in blood clotting. After an injury, clots normally protect the body by sealing off damaged blood vessels and preventing further blood loss.

The ADAMTS13 enzyme processes a large protein called von Willebrand factor, which also plays a role in clot formation. The unprocessed form of von Willebrand factor interacts easily with cell fragments called platelets, which circulate in the bloodstream and are essential for blood clotting. The factor helps platelets stick together and adhere to the walls of blood vessels, forming temporary clots. The ADAMTS13 enzyme cuts von Willebrand factor into smaller pieces. By processing von Willebrand factor in this way, the enzyme prevents it from triggering the formation of unnecessary blood clots.

Health Conditions Related to Genetic Changes

Thrombotic thrombocytopenic purpura

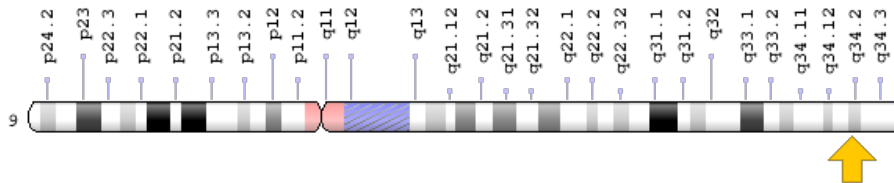
More than 70 mutations in the *ADAMTS13* gene have been reported in people with the familial form of thrombotic thrombocytopenic purpura. Most of these mutations change single protein building blocks (amino acids) in the ADAMTS13 enzyme. Other mutations lead to the production of an abnormally small version of the enzyme that cannot function properly.

Mutations in the *ADAMTS13* gene severely reduce the activity of the ADAMTS13 enzyme. As a result, von Willebrand factor is not processed normally in the bloodstream. If the factor is not cut into smaller fragments by the ADAMTS13 enzyme, it promotes the formation of abnormal clots throughout the body. The large, uncut version of von Willebrand factor induces platelets to stick together and adhere to the walls of blood vessels, even in the absence of injury. Additional factors such as pregnancy, diarrhea, surgery, and infection likely play a role in triggering abnormal clotting. Blood clots can block blood flow through small vessels, causing damage to the brain, kidneys, heart, and other organs. Abnormal clotting also causes other complications associated with thrombotic thrombocytopenic purpura.

Chromosomal Location

Cytogenetic Location: 9q34.2, which is the long (q) arm of chromosome 9 at position 34.2

Molecular Location: base pairs 133,414,339 to 133,459,403 on chromosome 9 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ADAM metallopeptidase with thrombospondin type 1 motif, 13
- ADAMTS-13
- ATS13_HUMAN
- C9orf8
- von Willebrand factor-cleaving protease
- vWF-cleaving protease
- vWF-CP
- VWFCP

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ADAMTS13%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- A DISINTEGRIN-LIKE AND METALLOPROTEASE WITH THROMBOSPONDIN TYPE 1 MOTIF, 13
<http://omim.org/entry/604134>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ADAMTS13.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ADAMTS13%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:1366
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:11093>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/11093>
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