ADAMTS13 gene

ADAM metallopeptidase with thrombospondin type 1 motif 13

Normal Function

The *ADAMTS13* gene provides instructions for making an enzyme that is involved in regulating 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. This protein is involved in the first step of blood clotting at the site of injury, which is to help cells called platelets stick together and attach to the walls of blood vessels, forming temporary clots. The ADAMTS13 enzyme cuts von Willebrand factor into smaller pieces to regulate its interaction with platelets. By processing von Willebrand factor in this way, the enzyme prevents it from triggering the formation of blood clots in normal circulation.

Health Conditions Related to Genetic Changes

Thrombotic thrombocytopenic purpura

More than 150 mutations in the *ADAMTS13* gene have been reported in people with the familial form of thrombotic thrombocytopenic purpura. This condition causes blood clots (thrombi) to form in small blood vessels throughout the body. These clots can cause serious medical problems if they block vessels and restrict blood flow to organs such as the brain, kidneys, and heart. Complications resulting from these clots can include neurological problems (such as personality changes, headaches, confusion, and slurred speech), fever, abnormal kidney function, abdominal pain, and heart problems.

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. These mutations 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 uncut version of von Willebrand factor is hyperactive and may induce platelets to stick together, even in the absence of injury, leading to the signs and symptoms of thrombotic thrombocytopenic purpura.

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

Tests Listed in the Genetic Testing Registry

- Tests of ADAMTS13 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=11093[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; ADAMTS13 (https://omim.org/entry/604134)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/11093)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ADAMTS13[gene])

References

- Levy GG, Nichols WC, Lian EC, Foroud T, McClintick JN, McGee BM, Yang AY, Siemieniak DR, Stark KR, Gruppo R, Sarode R, Shurin SB, Chandrasekaran V, StablerSP, Sabio H, Bouhassira EE, Upshaw JD Jr, Ginsburg D, Tsai HM. Mutations in amember of the ADAMTS gene family cause thrombotic thrombocytopenic purpura.Nature. 2001 Oct 4;413(6855):488-94. doi: 10.1038/35097008. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11586351)

- Tsai HM. Is severe deficiency of ADAMTS-13 specific for thrombotic thrombocytopenic purpura? Yes. J Thromb Haemost. 2003 Apr;1(4):625-31. doi:10.1046/j.1538-7836.2003.00169.x. No abstract available. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12871390)
- Tsai HM. Thrombotic thrombocytopenic purpura and the atypical hemolytic uremic syndrome: an update. Hematol Oncol Clin North Am. 2013 Jun;27(3):565-84. doi:10.1016/j.hoc.2013.02.006. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23714312)

Genomic Location

The *ADAMTS13* gene is found on chromosome 9 (https://medlineplus.gov/genetics/chromosome/9/).

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