VWF gene

von Willebrand factor

Normal Function

The *VWF* gene provides instructions for making a blood clotting protein called von Willebrand factor. This protein contains regions that attach (bind) to specific cells and proteins during the formation of a blood clot. After an injury, clots protect the body by sealing off damaged blood vessels and preventing further blood loss.

Von Willebrand factor is made within endothelial cells, which line the inside surface of blood vessels, and bone marrow cells. The factor is made of several identical subunits. To facilitate binding to various cells and proteins, these subunits are cut into smaller pieces by an enzyme called ADAMTS13. Von Willebrand factor helps platelets stick together and adhere to the walls of blood vessels at the site of a wound. These groups of platelets form temporary clots, plugging holes in blood vessel walls to help stop bleeding. Von Willebrand factor also carries another blood clotting protein, coagulation factor VIII, to the area of clot formation.

Health Conditions Related to Genetic Changes

Von Willebrand disease

More than 300 mutations in the *VWF* gene have been found to cause von Willebrand disease. Mutations in the *VWF* gene that reduce the amount of von Willebrand factor cause type 1 von Willebrand disease. People with type 1 von Willebrand disease have von Willebrand factor in their bloodstream, but at reduced amounts. Mutations that disrupt the function of the von Willebrand factor cause the four subtypes of type 2 von Willebrand disease. These mutations usually change one of the protein building blocks (amino acids) used to make von Willebrand factor, which can disrupt the factor's ability to bind to various cells and proteins needed to form a blood clot. Mutations that result in an abnormally short, nonfunctional von Willebrand factor generally cause the more severe type 3 von Willebrand disease. A reduction in the amount of von Willebrand factor or problems with its function slows the formation of blood clots, which causes the prolonged bleeding episodes seen in von Willebrand disease.

Other Names for This Gene

- coagulation factor VIII VWF

- F8VWF
- VWD

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of VWF (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7450[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28VWF%5BTI%5D%29+OR+%28von+Willebrand+factor%5BTI%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- VON WILLEBRAND FACTOR; VWF (https://omim.org/entry/613160)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7450)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=VWF[gene])

References

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Genomic Location

The *VWF* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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