GP9 gene

glycoprotein IX platelet

Normal Function

The *GP9* gene provides instructions for making a protein called glycoprotein IX (GPIX). This protein is one piece (subunit) of a protein complex called GPIb-IX-V, which plays a role in blood clotting. GPIb-IX-V is found on the surface of small cells called platelets, which circulate in blood and are an essential component of blood clots. The complex can attach (bind) to a protein called von Willebrand factor, fitting together like a lock and its key. Von Willebrand factor is found on the inside surface of blood vessels, particularly when there is an injury. Binding of the GPIb-IX-V complex to von Willebrand factor allows platelets to stick to the blood vessel wall at the site of the injury. These platelets form clots, plugging holes in the blood vessels to help stop bleeding.

To form the GPIb-IX-V complex, GPIX interacts with other protein subunits called GPIb-alpha, GPIb-beta, and GPV, each of which is produced from a different gene. GPIX is essential for assembly of the complex at the platelet surface and helps stabilize the complex once it is formed.

Health Conditions Related to Genetic Changes

Bernard-Soulier syndrome

At least 28 *GP9* gene mutations have been found to cause Bernard-Soulier syndrome, a condition characterized by a reduced number of platelets that are larger than normal (macrothrombocytopenia) and excessive bleeding. These mutations lead to production of an altered GPIX subunit that is likely broken down too soon or that cannot get to the platelet surface. Lack of this subunit on the surface of platelets prevents formation of the GPIb-IX-V complex. Without GPIb-IX-V, platelets cannot come together at the site of an injury to form a clot, leading to the bleeding problems associated with Bernard-Soulier syndrome.

Other Names for This Gene

- CD42a
- glycoprotein 9
- glycoprotein IX (platelet)

- GPIX
- platelet glycoprotein IX precursor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GP9 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2815[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GP9%5BTIAB%5D%29+OR+%28glycoprotein+IX%5BTIAB%5D%29+OR+%28GPIX%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GLYCOPROTEIN IX, PLATELET; GP9 (https://omim.org/entry/173515)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2815)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GP9[gene])

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Genomic Location

The *GP9* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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