GP1BA gene

glycoprotein Ib platelet subunit alpha

Normal Function

The *GP1BA* gene provides instructions for making a protein called glycoprotein Ib-alpha (GPIbα). 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, GPIbα interacts with other protein subunits called GPIb-beta, GPIX, and GPV, each of which is produced from a different gene. GPIbα is essential for assembly of the complex at the platelet surface. It is the piece of the complex that interacts with von Willebrand factor to trigger blood clotting. GPIbα also interacts with other blood clotting proteins to aid in other steps of the clotting process.

Health Conditions Related to Genetic Changes

Bernard-Soulier syndrome

At least 54 *GP1BA* 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. Some of these mutations lead to production of an altered GPIbα 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 mutations lead to production of a subunit that can form GPIb-IX-V complexes but cannot interact with von Willebrand factor, which also impairs the accumulation of platelets necessary for clotting.

Other disorders

At least six mutations in the *GP1BA* gene can cause another bleeding disorder called platelet-type von Willebrand disease (also known as pseudo-von Willebrand disease). This disorder is characterized by a reduced number of platelets in the blood (thrombocytopenia) and mild bleeding abnormalities. In contrast to mutations that cause Bernard-Soulier syndrome (described above), mutations involved in platelet-type von Willebrand disease lead to excessive binding of the GPIb-IX-V complex to von Willebrand factor. Because platelets containing the altered complex attach to von Willebrand factor without an injury to the blood vessel, fewer platelets are available for clot formation when an injury occurs, which leads to excessive bleeding in people with platelet-type von Willebrand disease.

Other Names for This Gene

- antigen CD42b-alpha
- BDPLT1
- BDPLT3
- BSS
- CD42B
- CD42b-alpha
- DBPLT3
- glycoprotein Ib (platelet), alpha polypeptide
- glycoprotein Ib platelet alpha subunit
- GP-Ib alpha
- GP1B
- GPIbA
- GPIbalpha
- platelet glycoprotein Ib alpha chain precursor
- platelet membrane glycoprotein 1b-alpha subunit
- VWDP

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GP1BA (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2811[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GP1BA%5BTIAB%5D%29+OR+%28glycoprotein+Ib+platelet+alpha+subunit%5BTIAB%5D%29%29+OR+%28%28CD42B%5BTIAB%5D%29+OR+%28GP-Ib+alpha%5BTIAB%5D%29+OR+%

28GP1B%5BTIAB%5D%29+OR+%28GPIbA%5BTIAB%5D%29+OR+%28GPIbalpha%5BTIAB%5D%29+OR+%28glycoprotein+Ib++,+alpha+polypeptide%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+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- VON WILLEBRAND DISEASE, PLATELET-TYPE; VWDP (https://omim.org/entry/177820)
- GLYCOPROTEIN Ib, PLATELET, ALPHA POLYPEPTIDE; GP1BA (https://omim.org/entry/606672)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2811)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GP1BA[gene])

References

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Genomic Location

The *GP1BA* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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