

GP1BA gene

glycoprotein Ib platelet subunit alpha

Normal Function

The *GP1BA* gene provides instructions for making a protein called glycoprotein Ib-alpha (GPIb α). 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, GPIb α interacts with other protein subunits called GPIb-beta, GPIX, and GPV, each of which is produced from a different gene. GPIb α 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. GPIb α 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 GPIb α 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\[geneid\]](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+lb+platelet+alpha+subunit%5BTIAB%5D%29%29+OR+%28%28CD42B%5BTIAB%5D%29+OR+%28GP-lb+alpha%5BTIAB%5D%29+OR+%>)

28GP1B%5BTIAB%5D%29+OR+%28GP1bA%5BTIAB%5D%29+OR+%28GP1balpha%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\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GP1BA[gene]))

References

- Cauwenberghs N, Vanhoorelbeke K, Vauterin S, Deckmyn H. Structural determinants within platelet glycoprotein Ib α involved in its binding to von Willebrand factor. *Platelets*. 2000 Nov;11(7):373-8. doi:10.1080/09537100020019157. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11132103>)
- Li R, Emsley J. The organizing principle of the platelet glycoprotein Ib-IX-V complex. *J Thromb Haemost*. 2013 Apr;11(4):605-14. doi: 10.1111/jth.12144. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23336709>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3696474/>)
- Othman M, Kaur H, Emsley J. Platelet-type von Willebrand disease: new insights into the molecular pathophysiology of a unique platelet defect. *Semin Thromb Hemost*. 2013 Sep;39(6):663-73. doi: 10.1055/s-0033-1353442. Epub 2013 Aug 11. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23934752>)
- Savoia A, Kunishima S, De Rocco D, Zieger B, Rand ML, Pujol-Moix N, Caliskan U, Tokgoz H, Pecci A, Noris P, Srivastava A, Ward C, Morel-Kopp MC, Alessi MC, Bellucci S, Beurrier P, de Maistre E, Favier R, Hezard N, Hurtaud-Roux MF, Latger-Cannard V, Lavenu-Bombled C, Proulle V, Meunier S, Negrier C, Nurden A, Randrianaivo H, Fabris F, Platokouki H, Rosenberg N, HadjKacem B, Heller PG, Karimi M, Balduini CL, Pastore A, Lanza F. Spectrum of the mutations in Bernard-Soulier syndrome. *Hum Mutat*. 2014 Sep;35(9):1033-45. doi:10.1002/humu.22607. Epub 2014 Jul 15. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24934643>)
- Schade AJ, Arya M, Gao S, Diz-Kucukkaya R, Anvari B, McIntire LV, Lopez JA, Dong JF. Cytoplasmic truncation of glycoprotein Ib α weakens its interaction with

von Willebrand factor and impairs cell adhesion. Biochemistry. 2003 Feb25;42(7):2245-51. doi: 10.1021/bi026549n. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12590614>)

Genomic Location

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

Last updated June 1, 2016