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
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.

**Chromosomal Location**

Cytogenetic Location: 17p13.2, which is the short (p) arm of chromosome 17 at position 13.2

Molecular Location: base pairs 4,932,277 to 4,935,023 on chromosome 17 (Homo sapiens Updated Annotation Release 109.20200228, GRCh38.p13) (NCBI)

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

Educational Resources
• Clinical Methods: The History, Physical, and Laboratory Examinations (third edition, 1990): Excessive Bleeding and Bruising, Basic Science
• Platelet-Vessel Wall Interactions in Hemostasis and Thrombosis (2010): Platelet Adhesion to Vascular Walls
  https://www.ncbi.nlm.nih.gov/books/NBK53456/

Scientific Articles on PubMed
• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GP1BA%5BTIAB%5D%29+OR+%28glycoprotein+Ib+alpha+subunit%5BTIAB%5D%29%29+OR+%28%28CD42B%5BTIAB%5D%29+OR+%28GP-Ib+alpha%5BTIAB%5D%29+OR+%28GP1BA%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
• GLYCOPROTEIN Ib, PLATELET, ALPHA POLYPEPTIDE
  http://omim.org/entry/606672
• PSEUDO-VON WILLEBRAND DISEASE
  http://omim.org/entry/177820

Research Resources
• Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GP1BA.html
• ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GP1BA%5Bgene%5D
• HGNC Gene Symbol Report
• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2811
Sources for This Summary

- OMIM: GLYCOPROTEIN Ib, PLATELET, ALPHA POLYPEPTIDE http://omim.org/entry/606672


Reviewed: June 2016
Published: April 28, 2020