

## GP6 gene

glycoprotein VI platelet

### Normal Function

The *GP6* gene provides instructions for making a protein called glycoprotein VI (GPVI). GPVI is a receptor protein that is embedded in the outer membrane of blood cells called platelets, which are an essential component of blood clots. Normally, blood clots protect the body after an injury by sealing off damaged blood vessels and preventing further blood loss.

Receptor proteins, like GPVI, have specific sites into which certain other proteins, called ligands, fit like keys into locks. Together, ligands and their receptors trigger signals that affect cell function. The main ligand for GPVI is a protein called collagen, which is found on blood vessel walls. In response to an injury that causes bleeding, the GPVI protein attaches (binds) to collagen, which begins clot formation and signals additional platelets to come together to increase the size of the clot. The GPVI protein can also bind to a protein called fibrin, which is the main protein that forms blood clots.

### Health Conditions Related to Genetic Changes

#### Glycoprotein VI deficiency

At least five mutations in the *GP6* gene have been found to cause glycoprotein VI deficiency, a bleeding disorder associated with a decreased ability to form blood clots. The mutations lead to the production of no GPVI protein; an abnormally short, nonfunctional GPVI protein; or a protein that is less able to bind to collagen. Without GPVI binding to collagen, platelets cannot come together efficiently at the site of an injury to form a clot, leading to an increased risk of nosebleeds, abnormally heavy or prolonged bleeding following minor injury or surgery, or other bleeding problems associated with glycoprotein VI deficiency.

### Other Names for This Gene

- glycoprotein 6
- GPVI
- GPVI collagen receptor
- platelet collagen receptor

- platelet glycoprotein VI
- platelet membrane glycoprotein VI

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of GP6 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51206\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51206[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GP6%5BTIAB%5D%29+OR+%28glycoprotein+VI+platelet%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%29%29%29>)

### Catalog of Genes and Diseases from OMIM

- GLYCOPROTEIN VI, PLATELET; GP6 (<https://omim.org/entry/605546>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/51206>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GP6\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GP6[gene]))

## References

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GP6 generates a truncated protein associated with a bleeding disorder in four Chilean families. J Thromb Haemost. 2013 Sep;11(9):1751-9. doi: 10.1111/jth.12334. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23815599>)

## **Genomic Location**

The *GP6* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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