

## **PLG gene**

plasminogen

### **Normal Function**

The *PLG* gene provides instructions for making a protein called plasminogen, which is produced in the liver. Enzymes called plasminogen activators convert plasminogen into the protein plasmin, which breaks down another protein called fibrin. Fibrin is the main protein involved in blood clots and is important for wound healing, creating the framework for normal tissue to grow back. Excess fibrin is broken down when no longer needed, and the new, more flexible normal tissue takes its place.

### **Health Conditions Related to Genetic Changes**

#### Congenital plasminogen deficiency

More than 50 mutations in the *PLG* gene have been identified in people with congenital plasminogen deficiency. This disorder is characterized by hard growths on the mucous membranes, which are the moist tissues that line body openings such as the eyelids and the inside of the mouth. Congenital plasminogen deficiency most often affects the conjunctiva, which are the mucous membranes that protect the white part of the eye (the sclera) and line the eyelids.

*PLG* gene mutations can decrease the amount of plasminogen produced, its function, or both. When the mutations affect plasminogen levels as well as the activity of the protein, affected individuals may be said to have type I congenital plasminogen deficiency, characterized by the growths previously described. People with *PLG* gene mutations that result in normal levels of plasminogen with reduced activity are said to have type II congenital plasminogen deficiency or dysplasminogenemia. This form of the condition often has no symptoms.

A reduction in functional plasminogen results in less plasmin to break down fibrin, leading to a buildup of fibrin. The excess fibrin and the resulting inflammation of the tissue result in the inflamed growths characteristic of congenital plasminogen deficiency.

It is unclear why the excess fibrin builds up in the mucous membranes but does not usually result in abnormal clots in the blood vessels (thromboses). Researchers suggest that other enzymes in the blood may also break down fibrin, helping to compensate for the reduced plasminogen levels.

## Other Names for This Gene

- DKFZp779M0222
- plasmin
- plasminogen isoform 1 precursor
- plasminogen isoform 2 precursor
- PLMN\_HUMAN

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of PLG ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5340\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5340[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PLG%5BTI%5D%29+OR+%28plasminogen%5BTI%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+360+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- PLASMINOGEN; PLG (<https://omim.org/entry/173350>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5340>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PLG\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PLG[gene]))

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## Genomic Location

The *PLG* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

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