# PIGV gene

phosphatidylinositol glycan anchor biosynthesis class V

## Normal Function

The *PIGV* gene provides instructions for making an enzyme called GPI mannosyltransferase 2. This enzyme takes part in a series of steps that produce a molecule called a glycosylphosphosphatidylinositol (GPI) anchor. Specifically, GPI mannosyltransferase 2 adds the second of three molecules of a complex sugar called mannose to the GPI anchor. This step takes place in the endoplasmic reticulum, which is a structure involved in protein processing and transport within cells. The complete GPI anchor attaches (binds) to various proteins in the endoplasmic reticulum. After the anchor and protein are bound, the anchor attaches itself to the outer surface of the cell membrane, ensuring that the protein will be available when it is needed.

## Health Conditions Related to Genetic Changes

Mabry syndrome

At least 14 mutations in the *PIGV* gene have been found to cause Mabry syndrome, a condition characterized by intellectual disability, distinctive facial features, increased levels of an enzyme called alkaline phosphatase in the blood (hyperphosphatasia), and other signs and symptoms. These mutations change single protein building blocks ( amino acids) in the GPI mannosyltransferase 2 enzyme. The altered protein is less able to add mannose to the forming GPI anchor. The incomplete GPI anchor cannot attach to proteins; without the anchor, the proteins cannot bind to the cell membrane and are released from the cell.

An enzyme called alkaline phosphatase is normally attached to a GPI anchor. However, when the anchor is impaired, alkaline phosphatase cannot be anchored to the cell membrane. Instead, alkaline phosphatase is released from the cell. This abnormal release of alkaline phosphatase is responsible for the hyperphosphatasia in Mabry syndrome. It is unclear how *PIGV* gene mutations lead to the other features of Mabry syndrome, but these signs and symptoms are likely due to a lack of proper GPI anchoring of proteins to cell membranes.

## Other Names for This Gene

- dol-P-Man dependent GPI mannosyltransferase

- FLJ20477
- GPI mannosyltransferase 2
- GPI mannosyltransferase II
- GPI-MT-II
- HPMRS1
- phosphatidylinositol glycan anchor biosynthesis, class V
- PIG-V
- PIGV_HUMAN

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PIGV (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55650[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PIGV%5BTIAB%5D%29+OR+%28PIG-V%29+OR+%28GPI+mannosyltransferase+2%29+OR+%28GPI+mannosyltransferase+II%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- PHOSPHATIDYLINOSITOL GLYCAN ANCHOR BIOSYNTHESIS CLASS V PROTEIN; PIGV (https://omim.org/entry/610274)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/55650)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PIGV[gene])

## References


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## Genomic Location

The *PIGV* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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