# PGAP2 gene

post-GPI attachment to proteins 2

## Normal Function

The *PGAP2* gene provides instructions for making a protein that modifies a molecule called a glycosylphosphophatidylinositol (GPI) anchor. The GPI anchor attaches (binds) to various proteins and then binds them to the outer surface of the cell membrane, ensuring that they are available when needed. The GPI anchor is made up of many different pieces and is assembled in a cell structure called the endoplasmic reticulum, which is involved in protein processing and transport. The anchor is then transferred to a different cell structure called the Golgi apparatus, which modifies newly produced enzymes and other proteins. In the Golgi apparatus, the PGAP2 protein assists in attaching a molecule called a saturated fatty acid to the anchor. This saturated fatty acid is likely needed to help transport and attach the anchor to the fat-rich cell membrane.

## Health Conditions Related to Genetic Changes

Mabry syndrome

At least five *PGAP2* gene mutations have been found to cause Mabry syndrome. The features of Mabry syndrome include 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 PGAP2 protein and probably reduce the activity of the protein. As a result, the PGAP2 protein cannot efficiently modify the GPI anchor, likely impairing the anchor's ability to attach itself and its associated protein to the cell membrane. GPI anchor-associated proteins that cannot attach to the cell membrane are released from the cell.

An enzyme called alkaline phosphatase is normally attached to the cell membrane by a GPI anchor. However, when the anchor is impaired, 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 *PGAP2* 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

- cell wall biogenesis 43 N-terminal homolog
- CWH43-N
- FGF receptor activating protein 1
- FGF receptor-activating protein 1
- FRAG1
- PGAP2_HUMAN
- post-GPI attachment to proteins factor 2

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PGAP2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=27315[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28PGAP2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- POST-GPI ATTACHMENT TO PROTEINS 2; PGAP2 (https://omim.org/entry/615187)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/27315)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PGAP2[gene])

## References


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## Genomic Location

The *PGAP2* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

**Last updated August 1, 2013**