

PIGO Gene

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phosphatidylinositol glycan anchor biosynthesis class O

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1. Introduction

The *PIGO* gene provides instructions for making one part of an enzyme called GPI ethanolamine phosphate transfer 3 (GPI-ET3). The other part of the GPI-ET3 enzyme is produced from a gene called *PIGF*. The GPI-ET3 enzyme is involved in a series of steps that produce a molecule called a glycosylphosphatidylinositol (GPI) anchor. Specifically, this enzyme adds a molecule of ethanolamine phosphate to the end of the forming 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; this process requires the ethanolamine phosphate at the end of the anchor. 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.

2. Health Conditions Related to Genetic Changes

2.1. Mabry syndrome

At least three mutations in the *PIGO* 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-ET3 enzyme. The altered protein is less able to add ethanolamine phosphate to the end of GPI anchors. 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 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 *PIGO* 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.

3. Other Names for This Gene

- FLJ00135
- GPI ethanolamine phosphate transferase 3
- phosphatidylinositol glycan anchor biosynthesis, class O
- phosphatidylinositol-glycan biosynthesis class O protein
- PIG-O
- PIGO_HUMAN

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