# GPI Gene

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## 1. Introduction

The *GPI* gene provides instructions for making an enzyme called glucose phosphate isomerase (GPI). This enzyme has two distinct functions based on its structure. When two GPI molecules form a complex (a homodimer), the enzyme plays a role in a critical energy-producing process known as glycolysis, also called the glycolytic pathway. During glycolysis, the simple sugar glucose is broken down to produce energy. Specifically, GPI is involved in the second step of the glycolytic pathway; in this step, a molecule called glucose-6-phosphate is converted to another molecule called fructose-6-phosphate.

When GPI remains a single molecule (a monomer) it is involved in the development and maintenance of nerve cells (neurons). In this context, it is often known as neuroleukin (NLK).

The monomer is also produced by cancer cells and functions to promote spreading (metastasis) of the cancer; in this context it is called autocrine motility factor (AMF).

## 2. Health Conditions Related to Genetic Changes

### 2.1.Glucose phosphate isomerase deficiency

More than 30 *GPI* gene mutations have been identified in people with glucose phosphate isomerase (GPI) deficiency. GPI deficiency is an inherited disorder that affects red blood cells, which carry oxygen to the body's tissues. People with this disorder have a condition known as chronic hemolytic anemia, in which red blood cells are broken down (undergo hemolysis) prematurely, resulting in a shortage of red blood cells (anemia). Some affected individuals also have neurological problems, including intellectual disability and difficulty with coordinating movements (ataxia).

Most of the mutations that cause GPI deficiency replace single protein building blocks (amino acids) in the GPI enzyme. Some of these mutations may result in a less stable homodimer, impairing the activity of the enzyme in the glycolytic pathway. The resulting imbalance of molecules involved in the glycolytic pathway eventually impairs the ability of red cells to maintain their structure, leading to hemolysis.

Other *GPI* gene mutations may cause the monomer to break down more easily, thereby interfering with its function in nerve cells. In addition, the shortage of monomers hinders homodimer formation, which impairs the glycolytic pathway. These mutations have been identified in individuals with GPI deficiency who have both hemolytic anemia and neurological problems.

## 3. Other Names for This Gene

- AMF
- autocrine motility factor
- G6PI_HUMAN
- glucose phosphate isomerase
- GNPI

- hexose monophosphate isomerase
- hexosephosphate isomerase
- neuroleukin
- NLK
- oxoisomerase
- PGI
- PHI
- phosphoglucose isomerase
- phosphohexomutase
- phosphohexose isomerase
- phosphosaccharomutase
- SA36
- sperm antigen 36

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