

Glucose phosphate isomerase (GPI) deficiency: is an enzyme of the glycolytic pathway necessary for normal RBC survival. GPI deficiency is the second most common red blood cell glycolytic enzymopathy after pyruvate kinase (PK) deficiency, and approximately 50 different cases have been described to date. GPI deficiency is an autosomal recessive genetic disorder associated with mild to severe chronic hemolytic anemia in homozygotes or compound heterozygotes.

What causes the disease and how common is it?

This is a genetic disease. It is linked to mutations of the GPI gene, encoding the GPI enzyme. These mutations lead to a total or reduced GPI activity. An individual can be heterozygous for the disorder (healthy carrier) when only one of the GPI genes is mutated, or homozygous or compound heterozygote (affected individual) when the two GPI genes are mutated.

What are the most frequent symptoms if I have the disease?

The common clinical manifestation is chronic haemolytic anaemia, and in very rare cases, it is associated with neurological dysfunction and granulocyte dysfunction .

Which treatment must I follow if I have the disease?

Supportive measures such as red cell transfusions in case of severe anaemia. In some cases spleen removal (splenectomy) may improve the anaemia and thereby patient's clinical situation.

What is the risk of passing the condition on to my children?

Two people who carry each one copy of one of the mutated GPI gene have a 25 percent risk of having a child affected by the disorder at each pregnancy. The risk of having a child who is a healthy carrier of the disorder is 50 percent at each pregnancy, and the risk that a child will not have the disorder and will not be a carrier is 25 percent. Ask for genetic counselling to get a complete explanation.