

**Triose phosphate isomerase deficiency -TPI-** TPI is a glycolytic pathway enzymopathy leading to haemolytic anaemia of variable degree which is always associated with severe and progressive neurological degradation, myopathy and increased susceptibility to infection. TPI deficiency is very rare and transmitted in an autosomal recessive mode.

#### **What causes the disease and how common is it?**

This is a genetic disease. It is linked to mutations of the TPI gene, encoding the TPI enzyme. These mutations lead to a reduced TPI activity in patient's red blood cells. An individual can be heterozygous for the disorder (healthy carrier) when only one of the TPI genes is mutated, or homozygous or compound heterozygous (affected individual) when the two TPI genes are mutated.

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

TPI deficiency is characterized by an early-onset of haemolytic anaemia with severe jaundice, during the neonatal period of life. Neurological involvement is progressive and starts between 6 and 30 months of age leading to progressive neurological dysfunction, cardiomyopathy and increased susceptibility to infection. Most affected individuals die in early childhood, before the age of six years.

#### **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. There is no treatment for preventing progressive neurological impairment or any other non-haematological clinical manifestation of the disease.

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

Two people who carry each one copy of one of the mutated TPI genes 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.