Phosphofructokinase deficiency -PFK-: PFK is a key regulatory enzyme in glycolysis. PFK deficiency is a genetic disorder that interferes with the ability of muscles to use carbohydrates (such as glucose) for energy. It is also known as Tarui's Disease. This enzymopathy has a autosomal recessive hereditary transmission.

What causes the disease and how common is it?

This is a genetic disease. It is linked to mutations of the PFK genes, encoding the PFK enzyme. These mutations lead to reduced PFK activity. An individual can be heterozygous for the disorder (healthy carrier) when only one of the PFK genes is mutated, or homozygous or compound heterozygote (affected individual) when the two PFK genes are mutated. This enzymopathy is specially frequent in some ethnical groups such as Japanese and Jewish.

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

A major symptom is muscle pain during intense exercise. This autosomal recessive disease is characterized by mild to severe chronic haemolytic anaemia associated with dark urines (myoglobinuria) , muscle weakness, cramps, and exercise intolerance.

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 but not muscle pain and other clinical manifestations due to the myopathy.

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

Two people who carry each one copy of one of the mutated PFK 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.

Author: Richard van Wijk

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