Hexokinase deficiency -HK-: HK deficiency is a very rare autosomal, recessively inherited enzymopathy associated with severe chronic haemolytic anaemia (CHA)

What causes the disease and how common is it?

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

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

Clinical manifestations of Hexokinase (HK) deficiency are very similar to those of pyruvate kinase (PK) deficiency but anaemia is, in general, more severe. This is due to the concomitant shortage of 2,3 DPG, a metabolite that facilitates tissue oxygenation.

Which treatment must I follow if I have the disease?

In cases of severe anaemia supportive measures and red cell transfusions are indicated. Spleen removal (splenectomy) may improve the anaemia in these cases 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 HK 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.

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