

Adenylate kinase deficiency -AK-: AK is an enzyme involved in red cell nucleotide metabolism and necessary for normal RBC survival. AK deficiency is an autosomal recessive enzymopathy. In some cases, the chronic haemolytic anaemia is accompanied by neurological disease.

What causes the disease and how common is it?

This is a genetic disease. It is linked to mutations of the AK gene, encoding the AK enzyme. These mutations lead to a reduced AK activity in red blood cells. This deficiency has been reported in 12 unrelated families distributed worldwide.

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

It is a rare deficiency that causes chronic haemolytic anaemia. In some patients with AK deficiency the anaemia is associated with neurological disorder.

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 AK 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.