

Aldolase deficiency -ALD-: ALD is an enzyme of the anaerobic glycolytic pathway necessary for normal RBC survival. Aldolase A deficiency is an autosomal recessive enzyme defect.

What causes the disease and how common is it?

This is an extremely rare genetic disease and only six cases have been reported. It is linked to mutations of the ALD gene, encoding the ALD enzyme. These mutations lead to reduced ALD activity. An individual can be heterozygous for the disorder (healthy carrier) when only one of the ALD genes is mutated, or homozygous or compound heterozygotes (affected individual) when the two ALD genes are mutated.

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

Deficiency of Aldolase A can lead to muscular disorders (myopathies) with exercise intolerance or breakdown of muscle fibres (rhabdomyolysis) associated with haemolytic anaemia. Psychomotor retardation or mental retardation have also been reported.

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 ALD 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.