Glutathione reductase deficiency -GR: GR deficiency is an autosomal recessive genetic disorder. Congenital GR deficiency is associated with acute haemolytic crisis after oxidant drugs of fava beans ingestion (favism). In the only three cases so far reported, favism is always associated with cataracts.

What causes the disease and how common is it?

This is a very rare genetic disease. It is linked to mutations of the GR gene, encoding the GR enzyme. These mutations lead to a reduction of GR activity. Hereditary transmission is autosomal recessive and an individual can be heterozygous for the disorder (healthy carrier with one gene mutated) and homozygous or compound heterozygote (affected individual with two genes mutated). Up to now only three different families with congenital GR deficiency have been reported.

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

Clinical manifestations of GR deficiency are very similar to that of glucose-6-phosphate dehydrogenase (G6PD) deficiency (favism and severe neonatal jaundice). Cataracts may also be present.

Which treatment must I follow if I have the disease?

The treatment is mainly preventive: avoiding oxidative drugs and fava beans ingestion. In cases of severe haemolytic crises red cell transfusions may be necessary.

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

Two people who carry each one copy of one of the mutated GR 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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