Glutathione synthetase deficiency (GS): GSH plays an important role in biological functions, including synthesis of proteins and DNA and anti-oxidants detoxification. GS deficiency is an autosomal recessive genetic defect associated with chronic haemolytic anaemia, neurological disorder and metabolic acidosis.

What causes the disease and how common is it?

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

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

This enzyme deficiency is characterized by chronic nonspherocytic haemolytic anaemia. In severe cases the hemolytic anemia is associated with urinary elimination of 5-oxoproline, metabolic acidosis, and central nervous system damage.

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 GS 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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