Haemoglobin C disease: is a disorder of haemoglobin, a major component of the red blood cells. This abnormal haemoglobin precipitates in red blood cells to form crystals. It is a benign disease.

What causes the disease and how common is it?

This is a genetic disease. It is linked to a mutation of the □-globin gene, encoding the ß-globin chain, one of the components of haemoglobin (Hb). An individual can be heterozygous for the disorder (Hb AC individuals) when only one of the globin genes is mutated, or homozygous (Hb CC individuals) when the two beta globin genes are affected.
It is a frequent disorder in people originating from West- and North-Africa. This is due to the fact that these areas were or are still infected with malaria and Hb C confers a relative protection against malaria.

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

Most people with haemoglobin C disease have mild haemolytic anaemia and a normal life expectancy. In adulthood they might present enlargement of the spleen, gall stones and retinopathy. Heterozygotes usually have no disease.

Which treatment must I follow if I have the disease?

Most often no particular treatment is requested. If some complications happen, patients must consult specialist doctors.

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

Two people who carry each one copy of the mutated gene (Hb AC individuals) have a 25 percent risk of having a child affected by the disorder (Hb CC) 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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