

Haemoglobin Lepore: is a disorder of haemoglobin, a major component of the red blood cells. This abnormal haemoglobin is produced at a reduced rate. It might be a benign disease or result in a severe anaemia. See also the term: β -thalassemic syndromes ».

What causes the disease and how common is it?

This is a genetic disease. It resulted from a "patchwork" between the delta and beta globin genes. An individual can be heterozygous for the disorder (Hb ALepore individuals) when only one globin gene is a "patchwork", homozygous (Hb LeporeLepore individuals) when the two globin genes are affected, or compound heterozygous for the disorder (Hb Lepore- β -thalassaemia) when two beta-globin genes bear a different anomaly.

Hb Lepore is produced at a reduced rate and this is why the disease is related to the " β -thalassemic syndromes ».

Hb Lepore has a worldwide distribution, but it is more frequent in people originating from the Mediterranean Basin.

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

People heterozygous for haemoglobin Lepore (Hb ALepore) have no symptoms and a normal life expectancy. At the examination of their blood, one can see that the red blood cells are smaller and paler. It should be distinguished from an iron deficiency.

Those homozygous (Hb LeporeLepore; a very rare situation) or compound heterozygous (Hb Lepore- β -thalassaemia) might suffer from a severe anaemia. They should be managed in a comprehensive multi-disciplinary program of care. See also the term " β -thalassemic syndromes ».

Which treatment must I follow if I have the disease?

Individuals heterozygous for the Hb Lepore request no particular treatment. If some complications happen, patients must consult specialist doctors.

In case of Hb Lepore- β -thalassaemia, transfusion therapy might be required. See also the term " β -thalassemic syndromes ».

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

Two people who carry each one copy of the modified have a 25 percent risk of having a child affected by the disorder (Hb LeporeLepore; Hb Lepore- β -thalassaemia) at each pregnancy. The risk of having a child who is a healthy carrier of the disorder (Hb Lepore or β -thalassaemia) 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.