Haemoglobin E: is a disorder of haemoglobin, a major component of the red blood cells. This abnormal haemoglobin is produced at a reduced rate. It is most often a benign disease. See also the term: "Beta-thalassaemia".

What causes the disease and how common is it?

This is a genetic disease. It is linked to a mutation of the β-globin gene, encoding the ß-globin chain, one of the components of haemoglobin (Hb). An individual can be heterozygous for the disorder (individuals HbAE) when only one of the globin genes is mutated, homozygous (individuals HbEE) when the two beta globin genes are affected, or compound heterozygous for the disorder (E-β-thalassaemia) when two different mutations are encountered on each β-globin gene. This abnormal haemoglobin is produced at a reduced rate and this is why the disease is related to the "β-thalassemic syndromes".
It is a frequent disorder in people originating from South-East Asia.

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

Heterozygous and homozygous people for haemoglobin E (Hb AE and Hb EE, respectively) 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 with an HbE-β-thalassaemia syndrome might suffer from a severe anaemia. They should be managed in a comprehensive multi-disciplinary program of care.

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. In case of Hb Eβ-thalassaemia, transfusion therapy might be required.

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

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

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