Beta-thalassaemia major (intermedia): is a disorder of haemoglobin, a major component of the red blood cells. Haemoglobin is produced at a reduced rate. It is a severe disease.

Cooley's anaemia

What causes the disease and how common is it?

This is a genetic disease. It is linked to a decreased expression of two of the β-globin genes, encoding the β-globin chains, one of the components of haemoglobin (Hb).
It is a frequent disorder in people originating from the Mediterranean Basin, Asia and not rare in West-Africa. This is due to the fact that these areas were or are still infected with malaria and β-thalassaemia confers a relative protection against malaria.

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

Patients with β-thalassaemia major also called Cooley's anaemia, are healthy until the age of around six months and then become very pale due to profound anaemia. Without treatment, a child grows slowly, presents bone deformations, …
Patients with β-thalassaemia intermedia have a moderate form of the disease.

Which treatment must I follow if I have the disease?

Most often, without blood transfusion a patient will die rapidly. The disease and the blood transfusions are also responsible of a body iron excess which must be "removed" by using "iron chelators". The disease can be cured by a bone marrow transplantation. However it is not without side effects and should be discussed with specialists in the field.

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

Two people who carry each one copy of the mutated gene (β-thalassaemia trait) have a 25 percent risk of having a child affected by a more severe disorder (β-thalassaemia major or Cooley's anaemia) 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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