Unstable Haemoglobin: is a disorder of haemoglobin, a major component of the red blood cells. This abnormal haemoglobin is unstable and the red blood cell lifespan is reduced; it gives anaemia. 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). Individuals are heterozygous for the disorder (only one of the globin genes is mutated).
It is a rare disorder with a worldwide distribution.

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

Most people with haemoglobin M have cyanosis (blue color of lips and nails bed of the fingers) and some have also anaemia, but a normal life expectancy. In adulthood they might present enlargement of the spleen and gallstones.

Which treatment must I follow if I have the disease?

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

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

If one member of a couple has the mutated gene (Unstable Hb), there is a 50 percent risk of having a child affected by the disorder (Unstable Hb) at each pregnancy. The risk of having a child who has not the disorder is also 50 percent at each pregnancy. Ask for genetic counselling to get a complete explanation.

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