

**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  $\alpha$ -globin gene, encoding the  $\beta$ -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.