Haemoglobin S and sickle cell disorders: are disorders of the haemoglobin, a major component of the red blood cells. Sickle cell disorders (SCD) are the consequence of the presence of an abnormal haemoglobin called haemoglobin S (Hb S). There are several forms of which the most frequent is due to haemoglobin S homozygosity, while compound heterozygosity lead to a more or less severe form: SC, SD-Punjab, SO-Arab, Sβ-thalassaemia.

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). An individual can be heterozygous for the disorder (Hb AS individuals) when only one of the globin genes is mutated, or homozygous (Hb SS individuals), or compound heterozygotes HbSC, HbSD, HbSO-Arab, HbS□-thalassaemia individuals) when the two beta globin genes are affected. It is a frequent disorder in people originating from Africa, Middle-East, India, and the Mediterranean Basin. This is due to the fact that these areas were or are still infected with malaria and Hb S confers a relative protection against malaria.

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

Sometimes red blood cells from patients with sickle cell disorders become sickle-shaped (crescent shaped), have difficulty passing through small blood vessels and are destroyed rapidly. This explains why people with a sickle cell disorder have anaemia (pallor) and jaundice (yellow color of the eyes); when the circulation is blocked by the sickle-shaped red blood cells, it results in pain episodes (arms, legs, chest and abdomen), tissue damages (spleen, lung, liver, kidney, …), stroke and priapism (painful prolonged erection), … Damage to the spleen makes sickle cell disease patients, especially young children, easily overwhelmed by certain bacterial infections.
Heterozygotes usually have no disease and a normal life expectancy.

Which treatment must I follow if I have the disease?

If early diagnosis is made, treatment to prevent infectious episodes is started soon after birth. Treatment of complications is very important, this is why, like all patients with a chronic disease, they should be managed in a comprehensive multi-disciplinary program of care.
The disease can be cured by 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 one of the mutated gene have a 25 percent risk of having a child affected by the disorder (Hb SS; Hb SC, Hb SD, …) 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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