

**Atransferrinemia:** Atransferrinemia is an autosomal recessive condition that leads to a strong reduction of transferrin, the protein that transports iron in the blood circulation. Affected subjects show severe anemia and iron overload.

#### **What causes the disease and how common is it?**

This is an extremely rare genetic disease. It is linked to mutations of the TF gene that encodes for transferrin, the plasma protein that transports iron. Most cases of atransferrinemia are indeed a hypotransferrinemia, since the full absence of transferrin is likely incompatible with life. Atransferrinemia has been reported in few families worldwide. It is estimated that the prevalence of this diseases is  $<1 / 1\,000\,000$ .

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

A severe microcytic-hypochromic anemia since birth with the development of iron overload of liver and other organs. External signs are pallor and fatigue. Atransferrinemia appears early in life being its age of onset in neonatal or infancy period. Laboratory values of transferrin are half-normal in carriers and very low in affected patients.

#### **Which treatment must I follow if I have the disease?**

Periodic infusions of normal plasma (which contains transferrin) or purified apotransferrin may allow the correction of anemia and a normal development, without iron overload. Treatment with chelating agents (such as deferoxamine), phlebotomy of both can be required to diminish tissue iron overload.

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

Atransferrinemia is an autosomal recessive disease. An individual can be heterozygous for the disorder (healthy carrier) when only one of the TF allele is mutated, or homozygous or compound heterozygous (affected individual) when two TF allelels are mutated.

A couple who carries each one a mutated TF allele has a 25 percent risk of having a child affected by the disorder 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 probability 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.