Iron- refractory iron deficiency Anemia (IRIDA).: Matriptase-2 is a serine protease encoded by TMPRSS6 gene, whose function is important for intestinal iron absorption. Deficiency of TMRRSS6 is an autosomal recessive disorder called iron refractory iron deficiency anemia (IRIDA).

What causes the disease and how common is it?

This is a rare genetic disease, linked to mutations of the TMPRSS6 gene, encoding the serine protease matriptase 2. The mutations lead to a reduced activity of matriptase-2 in hepatocytes and thus to an increased amount of the hormone hepcidin which inhibits intestinal iron absorption.

So far 41 cases from 24 families distributed world-wide have been described with IRIDA. The estimated prevalence of the disease is <1 / 1 000 000.

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

TMPRSS6 deficiency leads to microcytic hypochromic anaemia of moderate degree since birth due to defective iron absorption because of inappropriately high production of the iron hormone hepcidin. The disease is known as IRIDA (iron refractory iron deficiency anemia).

Which treatment must I follow if I have the disease?

The degree of anemia is refractory to oral iron treatment, but may partially respond to parenteral (i.v.) iron that should be administered especially during growth.

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

IRIDA is an autosomal recessive disease. An individual can be heterozygous for the disorder (healthy carrier) when only one of the TMPRSS6 allelel is mutated, or homozygous or compound heterozygous (affected individual) when two TMPRSS6 allelels are mutated.

A couple who carries each one a mutated TMPRSS6 allele have 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 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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