

Microcytic anemia with liver iron overload (DMT1-deficiency Anemia with iron overload): Divalent metal transporter 1 (DMT1) is a duodenal apical iron transporter encoded by the SLC11A2 gene. Mutations in SLC11A2 leads to an anemia-iron overload syndrome transmitted as an autosomal recessive trait.

What causes the disease and how common is it?

This is a rare genetic disease caused by mutations in the SLC11A2 gene, encoding the DMT1 iron transporter. DMT1 is a key mediator of iron absorption and iron transfer from endosomes into the cytosol of developing erythroid cells.

Four cases have been described in the literature. The exact prevalence of this disease it is not known but it is estimated to be <1 / 1 000 000.

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

DMT1 deficiency leads to severe microcytic hypochromic anaemia present from birth.

Which treatment must I follow if I have the disease?

All reported patients have been treated with erythrocyte transfusions, although these patients do not required continued regular transfusion like beta-thalassemia patients. In one case erythropoietin (EPO) treatment allowed transfusion independency. Erythrocyte transfusion contributes to iron overload, however so far there is no data in these patients on iron chelation therapy. Continuous oral iron supplementation raises haemoglobin values improving quality of life.

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

DMT1-deficiency Anemia is an autosomal recessive disease. An individual can be heterozygous for the disorder (healthy carrier) when only one of the SLC11A2 allele is mutated, or homozygous or compound heterozygous (affected individual) when two SLC11A2 allelels are mutated.

A couple who carries each one a mutated SLC11A2 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 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.