Non-syndromic congenital sideroblastic anemia (CSA):
SLC25A38-related CSA.

Non-syndromic congenital sideroblastic anemia (CSA) is characterized by anemia and iron overload of erythroid precursors and other organs and it is due to mutations of at least 3 genes (ALAS2, SLC25A38 and GLRX5). SLC25A38-related CSA is a autosomal recessive sideroblastic anemia due to mutations of the mitochondrial transporter SLC25A38.

What causes the disease and how common is it?

Non-syndromic CSA is a rare genetic disease. The second most common type of non-syndromic CSA is due to mutations in the SLC25A38 gene. The SLC25A38 gene encode for a putative glycine/aminolevulinic acid transporter, the first substrates of the heme biosynthetic pathway.
Other forms of non-syndromic CSA are due to mutations of the ALAS2 (XLSA) and GLRX5 genes.
SLC25A38-related CSA is a rare disorder with fewer than 30 unrelated probands described in the literature. Estimated prevalence it is not known.

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

Deficiency of the SLC25A38 mitochondrial transporter causes severe microcytic hypochromic anemia with mitochondria iron accumulation in the erythroid cells and formation of excessive “ring sideroblasts” in the bone marrow. Iron overload may develop in the liver and other organs.

Which treatment must I follow if I have the disease?

SLC25A38-related CSA do not respond to treatment with vitamin B6 (pyridoxine) and patients often need regular blood transfusions for normal development. Iron overload is treated with chelation therapy. Bone marrow transplantation has been successfully done in few patients.

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

SLC25A38-related CSA is an autosomal recessive disease. An individual can be heterozygous for the disorder (healthy carrier) when only one of the SLC25A38 allelel is mutated, or homozygous or compound heterozygous (affected individual) when two SLC25A38 allelels are mutated.
A couple who carries each one a mutated SLC25A38 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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Date: 2005-2008