Congenital dyserythropoietic anaemia type II (CDA II) is a disorder of blood cell production, particularly of the production of erythroblasts, which are the precursors of the red blood cells (RBCs)

What causes the disease and how common is it?

This is a genetic disease. It is linked to a mutation of a gene, which regulates the proliferation, maturation and division of erythroblasts. The disorder is rare and is known in many regions in the world. Due to its rarity, the correct diagnosis is often made late, and although infants and children are affected, is sometimes made not earlier than in adulthood.

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

Less normal red cells are produced, and in many cases in addition life span of the red cells, which is normally about 110 days, is also reduced. These results in anaemia of varying degree. Severe forms: anaemia may be severe enough to require regular blood transfusion in childhood. In most cases, the anaemia becomes less severe in adolescents or adulthood, and regular transfusions beyond childhood are very rarely necessary.
Almost all affected individuals have a chronic moderate anaemia, which does not impair life expectancy, but may impair ability on exertion. In some patients, quality of life and function ability will again be reduced in higher age, particularly when the functions of the heart or the lungs are impaired. Additional symptoms are yellow discoloration in the eyes and sometimes of the skin. The spleen becomes enlarged, although the enlargement remains without symptoms. Other possible consequences are ulcers or bulks of extramedullary erythropoiesis along the spine seen in x-ray thorax, which may cause difficulties of diagnosis.
In almost all affected patients, there is a life long increased uptake of iron from the normal nutrition. This results in iron overload, which mains long time without consequences, but may later damage internal organs such as the liver or the heart. Therefore, life long control of iron metabolism is needed, e. g. in yearly intervals.

Which treatment must I follow if I have the disease?

In many cases, no particular treatment is requested. Treatment should be implemented, if in severe cases regular transfusions are needed, or if the physical ability and quality of life is impaired. Until now, no medications has proven its efficiency. The only possibility to improve the situation is the removal of an enlarged spleen in severe cases. However, since the spleen has relevant functions for the immunity, the decision whether this operation is of benefit should be made by the paediatrician or internist with special experience in the field. He of she will take measures to minimize the risk of a severe bacterial infection, and all patients without a functional spleen should be aware of the risks and carry a medical alert card to inform the physicians about the risk of infections which need immediate antibiotic treatment.
Life long control of iron metabolism is needed, e. g. in yearly intervals.

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 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 counseling to get a complete explanation.

Author: Hermann Heimpel

Date: 2005-2008