Congenital dyserythropoietic anaemia type I (CDA I) 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 the gene(s), which regulate the proliferation and maturation of erythroblasts. The disorder is very rare and is known in many regions in the world.

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

Less normal red cells are produced, and in some cases in addition the life span of the red cells, which is normally about 120 days, is also reduced. These results in anaemia of varying degree. 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 functional ability will again be reduced in higher age, particularly when the function 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 leg ulcers or bulks of extramedullary erythropoiesis along the spine seen in x-ray of the thorax, which may cause difficulties of diagnosis.
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
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 severe forms the 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 only rarely necessary.
In cases which require regular blood transfusions, or when the physical ability and quality of life is impaired, treatment by Interferon alpha, a normal hormone of the body can normalize the blood counts. This alleviates symptoms and prohibits further iron uptake. If indicated, this treatment has to be used for long times, possibly life long, with weekly or biweekly injections. This type of treatment has to be controlled by a specialist of paediatrics or internal medicine, who has experience with the treatment of chronic anaemias.

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.

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