

Congenital dyserythropoietic anemia type III (CDA III) is a very rare condition which has been observed in very few families in the world.

What causes the disease and how common is it?

This is a genetic disease, with two large families in North-Sweden and California.

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 110 days, is also reduced. This results in an moderate or light anaemia. Additional symptoms may be 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 thorax, which may cause difficulties of diagnosis.

Which treatment must I follow if I have the disease?

Usually, no special treatment is needed. The patient should be seen by a specialist of paediatrics or internal medicine in suitable intervals, and also by an ophthalmologist since changes endangering the vision may develop. .

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

If one member of a couple has the mutated gene, there is a 50 percent risk of having a child affected by the disorder at each pregnancy. The risk of having a child who has not the disorder is also 50 percent at each pregnancy. Ask for genetic counseling to get a complete explanation.