Other types of congenitale dyserythropoetic Anemia

What causes the disease and how common is it?

This is a genetic disease, with single families in many countries.

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 anemia of varying degree, with frequent impairment of physical ability. In severe cases regular blood transfusions may be needed. Additional symptoms may be yellow discoloration in the eyes and sometimes of the skin, and enlargement of the spleen. 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?

Unfortunately, no standard treatment is known. Removal of the spleen may be considered if the spleen is enlarged. The decision should be made by a specialist in cooperation with an expert centre. The patient should be seen by a specialist of paediatrics or internal medicine in suitable 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. However, the pattern of heredity may be different, and you should ask for genetic counseling to get a complete explanation.

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Date: 2005-2008