Phosphoglycerate kinase deficiency -PGK-: PGK is a key enzyme of the glycolytic pathway, involved in the production of ATP (energy). The PGK gene coding for this enzyme is located on the X-chromosome. PGK deficiency is a cause of chronic haemolytic anaemia and myoglobinuria.

What causes the disease and how common is it?

This deficiency is very rare, and only 26 unrelated families have been reported so far.

Since the PGK is a X-chromosome-linked mutation, hemizygous males and homozygous or compound heterozygotes females have reduced enzyme activity. Heterozygous females show variable enzyme expression depending on the pattern of X-chromosomes inactivation (lyonization).

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

PGK deficiency is characterized by chronic haemolytic anaemia (often fully compensated), central nervous system dysfunction (neurological disorder), and myopathy (muscle weakness and pain) with recurrent myoglobinuria (dark urine).

Which treatment must I follow if I have the disease?

In cases of severe chronic anaemia, regular blood transfusions are required. Spleen removal (splenectomy) may improve the clinical status in these cases.

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

Since this is a genetic disease linked to X chromosome, there are different parental combinations that may lead to an affected child, as following:

Parents	Children
Mother: Healthy carrier Father: Healthy	Female: 50% Healthy /50% Healthy carrier Male: 50% Healthy/ 50% Affected
Mother: Healthy Father: Affected	Female: 50% Healthy /50% Healthy carrier Male: 100% Healthy
Mother: Healthy carrier Father: Affected	Female: 50% Healthy carrier/50% Affected Male: 50% Healthy/ 50% Affected
Mother: Affected Father: Affected	Female: 100% Affected Male: 100% Affected

Author: Richard van Wijk

Date: 2005-2008