

Pyrimidine 5 nucleotidase deficiency -P5'N-1-: P5'N-1 is an enzyme involved in the catabolism of ribonucleic acid (RNA) from young RBCs (reticulocytes). P5'N-1 deficiency is an autosomal recessive disorder characterized by chronic haemolytic anaemia associated with prominent RBC basophilic stippling, which is very useful in diagnosing this enzyme deficiency. P5'N-1 deficiency is also implicated in the anaemia of lead poisoning and thalassaemia with basophilic stippling.

What causes the disease and how common is it?

P5'N-1 deficiency is the third RBC enzymopathy in frequency after PK deficiency and causes chronic nonspherocytic haemolytic anaemia. This enzymopathy is due to a mutation in the gene P5'N. An individual can be heterozygous for the disorder (healthy carrier) when only one gene is mutated, or homozygous or compound heterozygotes (affected individual) when the two genes are mutated.

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

Patients with P5'N-1 deficiency suffer from a long-life chronic haemolytic anaemia. The phenotype is usually relatively mild.

Which treatment must I follow if I have the disease?

Supportive measures such as red cell transfusions in case of severe anaemia. In some cases spleen removal (splenectomy) may improve the anaemia and thereby patient's clinical situation.

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

Two people who carry each one copy of one of the mutated P5'N-1 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 counselling to get a complete explanation.