



Glucose-6-phosphate dehydrogenase deficiency -G6PD: G6PD is the most frequent enzymopathy in human beings. G6PD is essential for the maintenance of adequate amounts of reduced glutathione (GSH) that prevents RBC from oxidative damage. This enzymopathy is symptomless until the patient is exposed to oxidant substances such as some drugs (for example anti-malarial medication) or fava beans (favism)

What causes the disease and how common is it?

G6PD deficiency is caused by an X-chromosome-linked mutation. Hemizygous man and homozygous or compound heterozygotes woman have reduced enzyme activity; whereas heterozygous woman have a variable enzyme expression depending on the pattern of X-chromosomes inactivation (lyonization). It is highly prevalent in several ethnical groups (Africans, Asians and Mediterraneans) where it confers a relative protection against malaria. G6PD deficiency is the most common human enzyme defect, affecting more than 400 million people worldwide.

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

Fortunately, most G6PD-deficient individuals are asymptomatic throughout their life, and unaware of their status. The most frequent clinical manifestations of G6PD deficiency are neonatal jaundice, and acute haemolytic anaemia, which is usually triggered by an exogenous agent. Some G6PD variants cause chronic haemolysis.

The illness generally manifests as acute haemolysis, which usually arises when red blood cells are challenged by oxidative stress, triggered by agents such as drugs, infection, or the ingestion of fava beans (favism). Acute haemolysis in G6PD deficiency, it is characterized by fatigue, back pain, anaemia, and jaundice. Several clinical disorders, such as diabetes, myocardial infarction or intense physical exercise have been reported to precipitate haemolysis in G6PD-deficient individuals; however, coexisting infection or oxidant drug exposure can be the underlying cause in these situations. In any case, G6PD deficiency does not seem to affect life expectancy, quality of life, or the activity of affected individuals.

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?

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

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