Glucose-6-phosphate dehydrogenase deficiency

Glucose-6-phosphate dehydrogenase deficiency (G6PDD) is one of the most common enzymatic defects worldwide. G6PD deficiency increases the sensitivity of erythrocyt to oxidative stress. It is clinically manifested by neonatal jaundice, acute hemolysis and, more rarely, chronic hemolytic anemia. People with this disease can also be asymptomatic.

Glucose-6-phosphate dehydrogenase deficiency:

Most patients with severe variants of G6PD develop favism or fabism , which is a predisposition to acute hemolysis after eating broad beans *(Vicia faba)* . So people with favism always have a G6PD deficiency, but not everyone with a G6PD deficiency has favism.

It is an X-linked inherited disease that occurs mainly in Africa, Asia, the Mediterranean and the Middle East. The number of people affected is estimated at 400 million people. Different types of genetic mutation in the G6PD gene are known (Xq28, OMIM: 305900 (https://omim.org/entry/305900)) responsible for different types of G6PD with different severe clinical manifestations.

Pathophysiology

Glucose-6-phosphate dehydrogenase (G6PD) catalyzes the reduction of NADP+ to NADPH in the pentose cycle . NADPH protects cells from oxidative stress . Because erythrocytes cannot produce NADPH in any other way, they are more sensitive to oxidative stress compared to other cells. As a result of oxidative stress, the cellular structure of erythrocytes changes, hemoglobin precipitates to form Heinz bodies (denatured hemoglobin), which subsequently causes the erythrocytes to break down (hemolysis).

Because NADPH is involved in respiratory burst of phages (specifically, reactions catalyzed by NADPH-phagosome oxidase), NADPH deficiency also leads to immunodeficiency. The ability of phagocytes to destroy absorbed material is reduced.

The clinical picture

There are different variants of G6PD deficiency, which have different severe clinical manifestations. Total G6PD deficiency is incompatible with life.

Neonatal jaundice

Newborns with G6PD (boys and homozygous girls) have a higher prevalence of hyperbilirubinemia, which usually develops within the first 24 hours of life, and may require phototherapy or exchange transfusion to prevent kernicterus.

Acute hemolysis

Acute hemolysis can be caused by infection, ingestion of Vicia faba beans (formerly vetch bob, formerly broad beans), use of certain drugs (eg aspirin, antibiotics - nitrofurantoin, sulfamethoxazole, chloramphenicol, ciprofloxacin; antimalarials and others) and contact with some substances (naphthalene). Acute haemolysis may be accompanied by back or abdominal pain and secondary jaundice due to an increase in unconjugated bilirubin levels

Acute hemolysis may be accompanied by transient splenomegaly a hemoglobinuria. Hemolysis typically occurs 24 to 72 hours after ingestion and resolves within 4 to 7 days. Rarely, hemolysis is so severe that it requires a blood transfusion.

Laboratory picture:

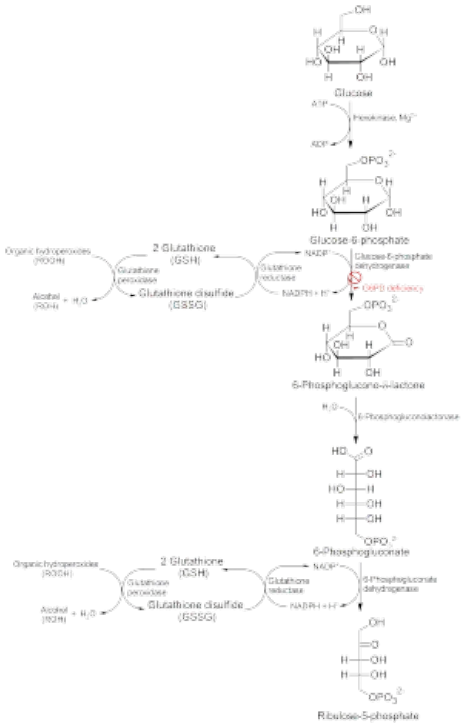
- Blood count – mild to severe anemia;
- Reticulocyte count – increased 4 to 7 days after hemolysis;
- Blood smear – Heinz bodies;
- Haptoglobin – decreased level;
- Liver tests – increased levels of indirect bilirubin;
- Coombs test – negative.

Diagnosis

G6PD deficiency is demonstrated by fluorescence assay or quantitative venous blood spectrophotometry.

Treatment

There is no causal treatment. Avoiding oxidative stressors is one of the preventive measures for acute hemolysis.



Pentose cycle disorder due to G6PD deficiency

Links

External links

- G6PDDeficiency.org (http://g6pddeficiency.org/wp/)
- Associazione Italiana Favismo (anglicky, německy) (https://www.g6pd.org/en/G6PDDeficiency.aspx)

- YOUNGSTER, Ilan. *Rare Diseases : Glucose-6-Phosphate Dehydrogenase Deficiency* [online]. [cit. 2022]. <https://rarediseases.org/rare-diseases/glucose-6-phosphate-dehydrogenase-deficiency/>.