





Glucose-6 phosphate dehydrogenase deficiency

Presentation by:

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Objective:

Explain glucose-6 phosphate dehydrogenase deficiency



Glucose-6 phosphate dehydrogenase deficiency.

- ✓ Glucose 6-phosphate dehydrogenase (G6PD) deficiency is an inherited disease characterized by hemolytic anemia caused by the inability to detoxify oxidizing agents.
- **✓** G6PD deficiency is the most common disease-producing enzyme abnormality in humans, affecting more than 200 million individuals worldwide.

- ✓ This deficiency has the highest prevalence in the Middle East, tropical Africa and Asia, and parts of the Mediterranean.
- ✓ G6PD deficiency is in fact, a family of deficiencies caused by more than 400 different mutations in the gene coding for G6PD.
- **✓** Only some of these mutations cause clinical symptoms.

- ✓ The life span of many individuals with G6PD deficiency is somewhat shortened as a result of complications arising from chronic hemolysis.
- ✓ This slightly negative effect of G6PD deficiency has been balanced in evolution by an advantage in increased resistance to falciparum malaria shown by female carriers of the mutation.

A. Role of G6PD in red blood cells:

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Glucose-6-phosphate
           G-6-P-D enzyme
6- Phosphoguconate + NADPH+H
   In case of G-6-P-D
            Leads to
      Inadequate supply of
          NADPH
            Leads to
      Reduced level of Glutathione
      Glutathione protects
         Hb oxidation
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B. Precipitating factors in G6PD deficiency:

- 1. Oxidant drugs
- 2. Favism
- 3. Infection
- 4. Neonatal jaundice

1. Oxidant drugs:

Commonly used drugs that produce hemolytic anemia in patients with *G6PD* deficiency are best remembered from the mnemonic **AAA** Antibiotics (for example, sulfamethoxazole methoxazole and chloramphenicol), Antimalarials (for example, primaquine but not quinine), and Antipyretics (for example, acetanilide but not acetaminophen).

2. Favism:

Some forms of *G6PD* deficiency, for example the Mediterranean variant, are particularly susceptible to the hemolytic effect of the fava bean, a dietary staple in the Mediterranean region. Favism, the hemolytic effect of ingesting fava beans, is not observed in all individuals with *G6PD* deficiency, but all patients with favism have *G6PD* deficiency.

3. Infection:

infection is the most common precipitating factor of hemolysis in *G6PD* deficiency. The inflammatory response to infection results in the generation of free radicals in macrophages, which can diffuse into the red blood cells and cause oxidative damage.

4. Neonatal jaundice:

Babies with *G6PD* deficiency may experience neonatal jaundice appearing one to four days after birth. The jaundice, which may be severe, results from impaired hepatic catabolism of heme or increased production of bilirubin.

