G6PD DEFICIENCY

By

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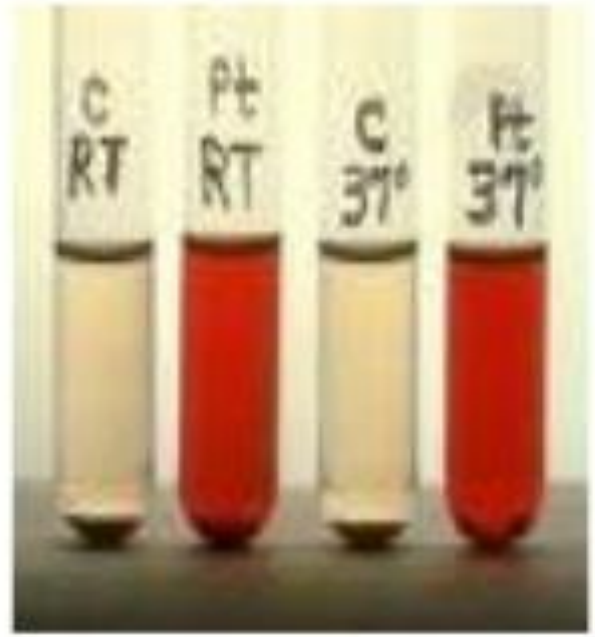
A ?? year-old boy with pallor (anemia) and red urine)(hemoglobinuria

Child 3 yrs old with sudden pallor , red urine and abdominal pain for 1 day before . admission

HISTORY OF FALAFEL



- ***What are characteristics of hemolytic anemia?***

  Hemolytic anemia is a state of decreased red blood cells survival. Patients with hemolytic anemia present with pallor, increased reticulocyte count which reflects bone marrow response and indirect hyperbilirubinemia which resulls from a breakdown of red blood cells.

Hemolytic anemia

- Cellular defects
  - Membrane defects: spherocytosis……
  - Enzyme deficiencies: G6PD deficiency….
  - Hemoblobin abnormalities: HbS, HbC…..
- Extracellular defects
  - Autoimmune
  - Fragmentation hemolysis: DIC, HUS….
  - Hypersplenism
  - Plasma factors: liver disease, infection…

G6PD DEFICIENCY

- X linked (mainly boys), less common AR
- Mediterranean countries , Arabic gulf countries, USA, china

- Pentose pathway (G6PD) ----→NADPH to prevent oxidative damage of RBC

Background on G6PD

- G6PD is an enzyme in the pentose phosphate pathway
- Converts NADP+ to NADPH
- G6PD deficiency is a sex-linked genetic disorders, with full expression in males
- Persons who are G6PD deficient are at increased risk for experiencing hemolytic anemia when taking FAVA

G6PD Genetic Variants

- (A -) Variant affects approximately 10% of African Americans
  - enzyme usually >10% normal

- (B -) Variant (MED) is the most common type affecting people from Eastern Mediterranean
  - Enzyme usually <10% of normal

G6PD deficiency

- Clinical manifestation
  - Neonatal jaundice
  - Acute hemolytic anemia
  - Chronic hemolytic anemia

G6PD deficiency-Acute hemolytic anemia

- Lab findings:
  - Anemia: moderate to extremely severe (Hgb: 2.5 g/dl)
  - PB smear: normocytic, normochromic anisocytosis, poikilocytosis bite cells
  - ↑Reticulocyte count: may reach 30%
  - ↓Haptoglobin

G6PD deficiency-Acute hemolytic anemia

- Lab findings:
  - Free hemoglobin in plasma (severe case)
  - ↑ WBC: granulocyte predominant
  - Plt: normal, ↑, ↓
  - ↑ LDH
  - ↑ unconjugated bilirubin
    liver enzyme: usually normal
  - Hemoglobinuria, dark urine test(+)

Treatment

- Hospitalization
- Packed RBC transfer till hemoglobin no more decrease and start to increase spontaneously
- Do G6PD enzyme assay after 4-6 weeks (because immature RBC still have the enzyme so pseudo resut)

FOOD, Drugs and Chemicals That Should Be Avoided by Persons With G6PD Deficiency

- 1- fava beans (alfoole)

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G6PDD

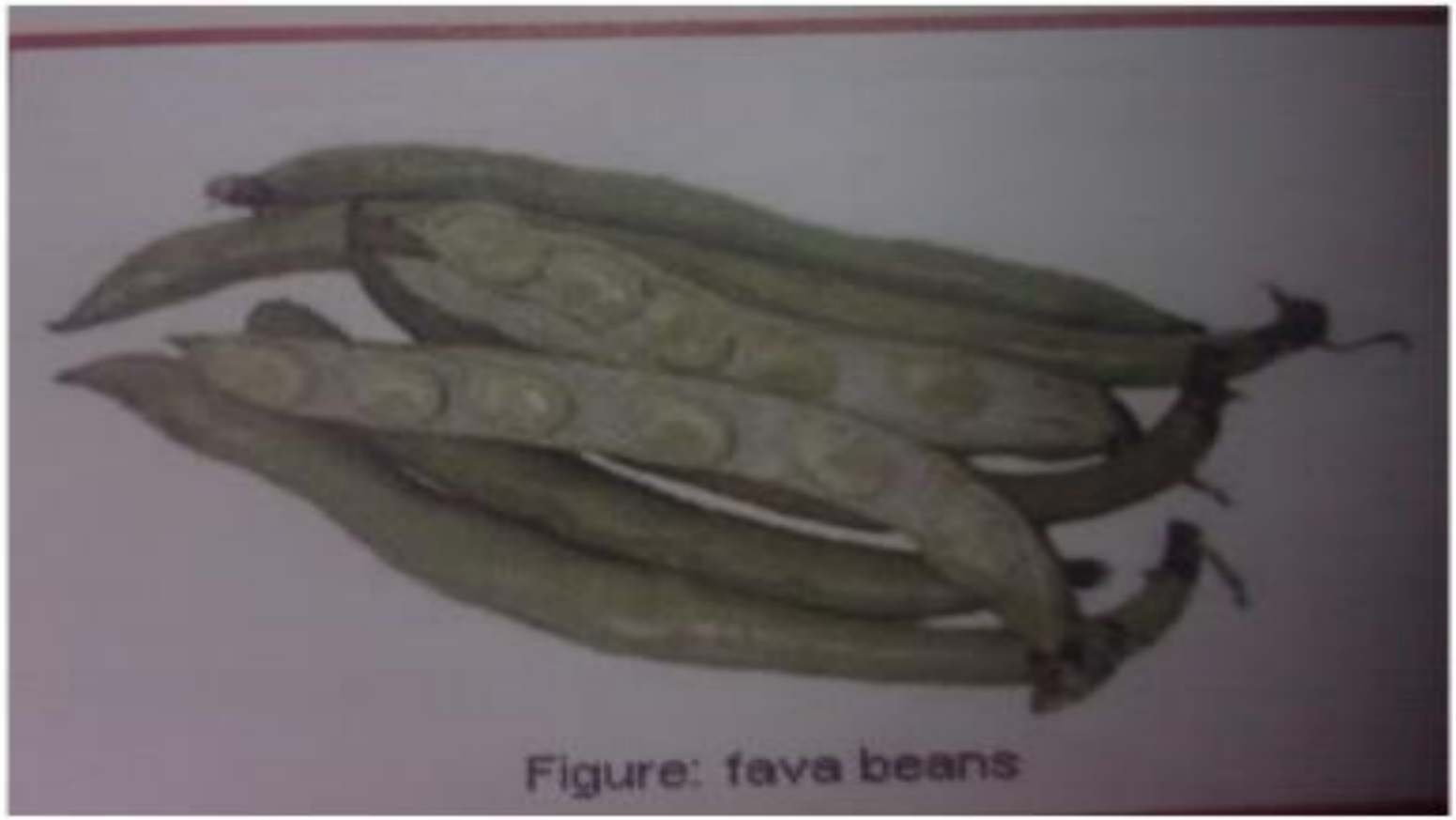
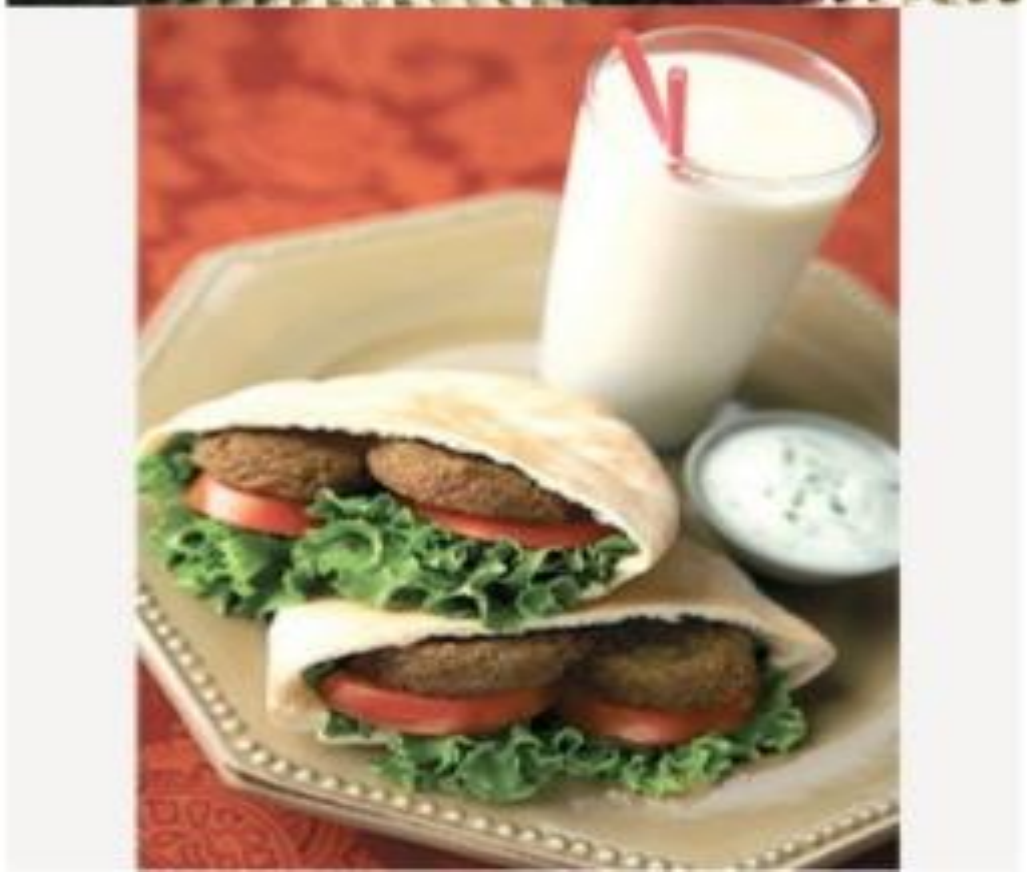


Figure: fava beans



FOOD, Drugs and Chemicals That Should Be Avoided by Persons With G6PD Deficiency

- 2- drugs:
- Antipyretics: except paracetamol
- Sulfa drugs
- Antimalarias
- Nitrofurans
- Antibiotics: chloramphenical
- Synthetic Vit K
- Infections : viral diseases as hepatitis,
- Diabetic ketoacidosis : DKA
- Chemicals : Methylene blue, naphthalene, benzene