G6PD deficiency

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The Red Cell

- Average life span
  - Adult: 120 days
  - Neonate: 60-90 days
  - Premature infants: 40-60 days

- Hemolytic anemias = reduced red cell life span

Hemolytic anemias

- Corpuscular (Intrinsic) or Extracorpuscular(Extrinsic)
- Acquired or hereditary

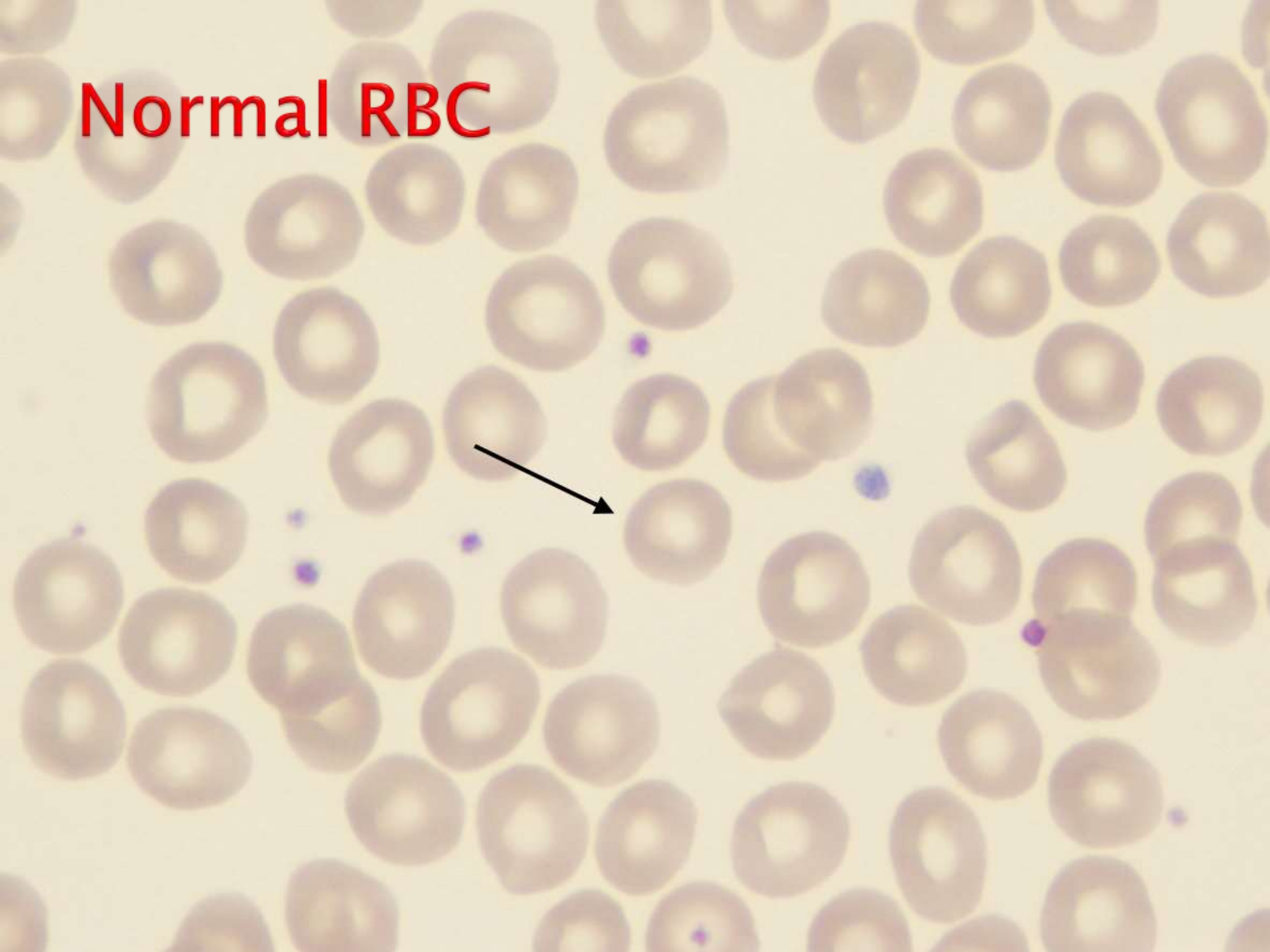
Classification of Hemolytic anemias

- **Corpuscular**
  - RBC membrane defects
    - HS, HE
  - Enzyme defects
    - PK, G6PD
  - Hemoglobin defects
    - Heme:
    - Globin: 1) quantitative: thalassemia
      2) qualitative: Sickle cell
- **Extracorpuscular**
  - Immune (Isoimmune, Autoimmune, Alloimmune)
  - Nonimmune

- Extracorpuscular
  - Immune (Isoimmune, Autoimmune, Alloimmune)
  - Nonimmune

- Autoimmune: Idiopathic, Secondary
- Nonimmune: Idiopathic, Secondary

Normal RBC



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Enzyme defects

G6PD Deficiency

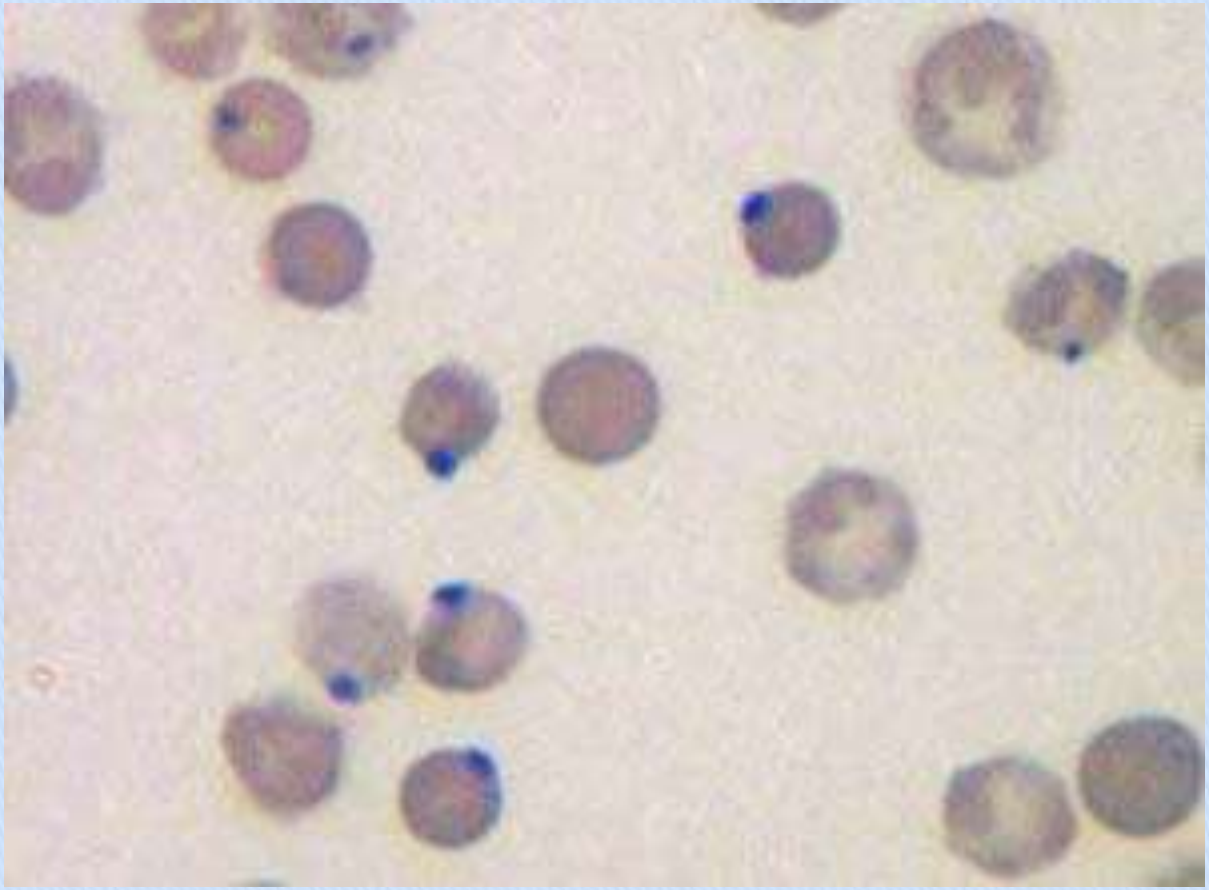
- MOST COMMON HUMAN ENZYME DEFECT
- Affecting 400-500 million people worldwide
- X-lined recessive, different variants of the disease
- Enzyme deficient red blood cells
- Higher prevalence in Mediterranean's, Blacks
- DECREASE IN GLUTATHIONE LEVELS

G6PD DEFICIENCY

- Enzyme deficient RBC are unable to defend against oxidant stress (infection, drugs) and form Heinz bodies (denatured hemoglobin) which are phagocytosed by splenic macrophages, creating “bites” on cells

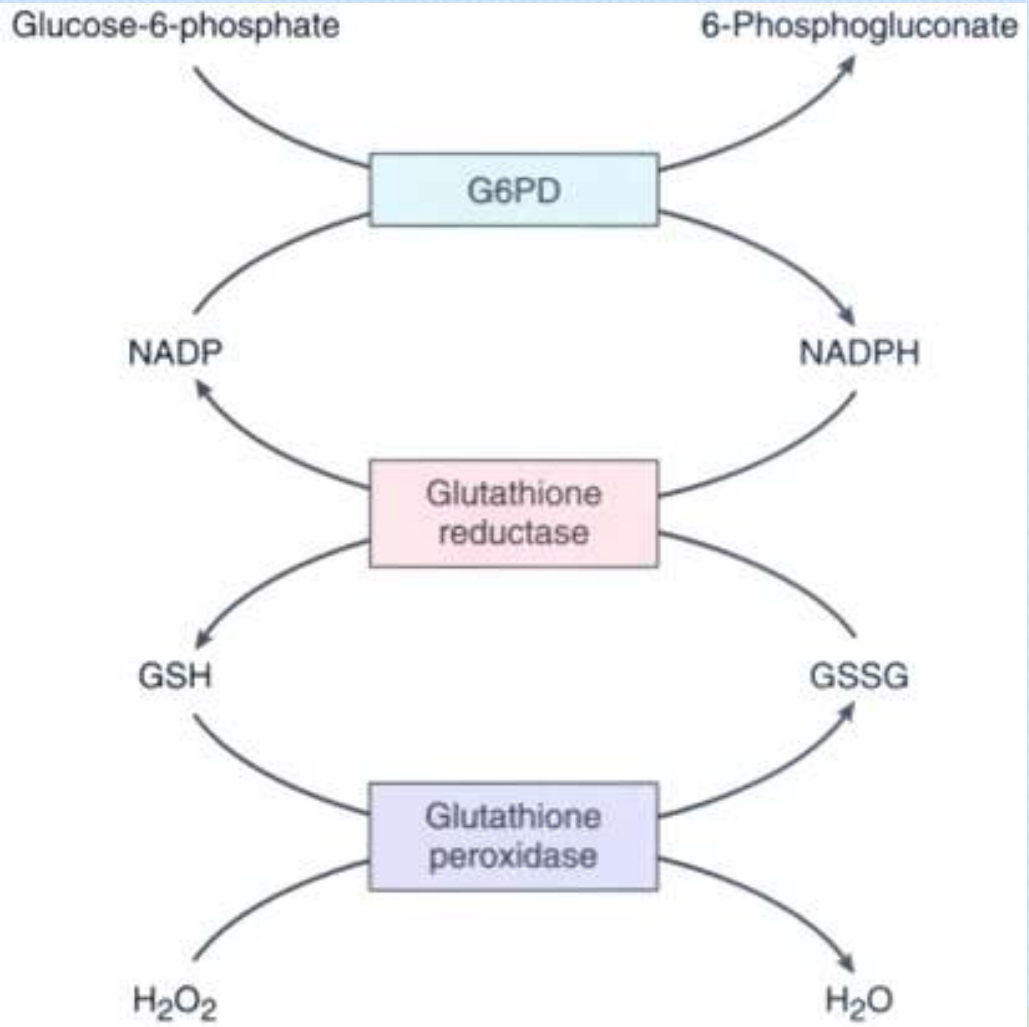
- Heinz bodies appear as granules in RBC’s

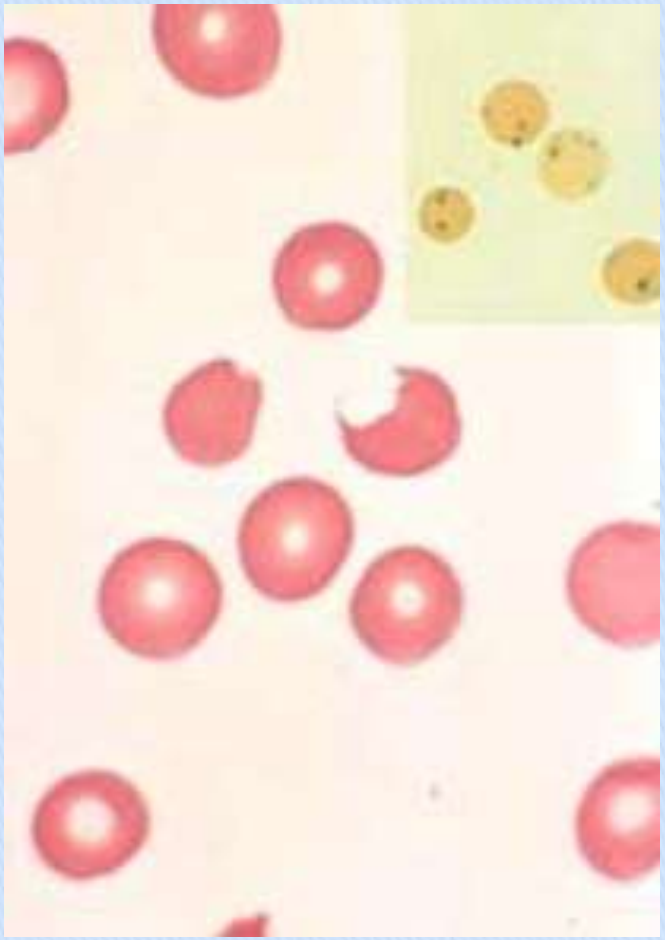
HEINZ bodies



Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency

- A^- and Mediterranean are most significant types





G6PD DEFICIENCY

Clinical manifestations

- Neonatal Jaundice
- Acute hemolytic anemia
- Chronic nonspherositic hemolytic anemia

G6PD DEFICIENCY

Acute hemolytic crisis

- Can be triggered by

  - Foods (fava beans)
  - Oxidant substances drugs
  - Infections
  - DKA

AGENTS PRECIPITATING HEMOLYSIS IN G6PD DEFICIENCY

- Antibacterials
- Sulfonamides
- Dapsone
- TMP-SMS
- Nalidixicacid
- Chloramphenicol
- Nitrofurantoin
- Antimalarials
- Primaquine
- Pamaquine
- Chloroquine
- Quinacrine

- Others
- Acetanilide
- VitaminK analogs
- Methyleneblue
- Probenecid
- ASA
- Phenazopyridine
- CHEMICALS
- Phenylhydrazine
- Benzene
- Naphthalene
- ILLNESS
- DKA
- Hepatitis
- Sepsis

TABLE 17-3 Drugs That Can Trigger Hemolysis in G6PD-Deficient Children*

Category of Drug	Definite Risk	Possible Risk
Antimalarials	Primaquine Dapsone-containing combinations†	Chloroquine Quinine
Analgesics	Acetanilid	Aspirin
Sulfonamides/ sulfones	Sulfamethoxazole/ co-trimoxazole Dapsone†	Sulfasalazine Sulfadiazine
Quinolones	Nalidixic acid Ciprofloxacin Norfloxacin Moxifloxacin Ofloxacin	
Other antimicrobials	Nitrofurantoin Methylene blue	Chloramphenicol
Other	Niridazole	Vitamin K Rasburicase Ascorbic acid Glibenclamide

G 6 P D deficiency

Clinically

- Most individuals with G6PD deficiency are asymptomatic
- Usually no evidence of hemolysis is apparent until 24-48 hours after the patient has ingested a substance which has oxidant properties

- Acute intravascular hemolysis can occur:
  - Hemoglobinuria
  - Dark urine
  - Hemoglobinemia
  - Anemia
  - Jaundice

G 6 P D deficiency

Diagnosis

- History

- Lab Tests: CBC, PBS, Retic, Na, K, BUN, Cr , AST, ALT, Bili, LDH, BG, Rh, Direct Coombs, G6PD, U/A

- Low G6PD activity in RBC
- G6PD assay best done when not in acute crisis as retic count is high

G 6 P D deficiency

Treatment: SUPPORTIVE , PREVENTATIVE

1. Hydration
2. Indications for Red blood cell transfusion
3. Education of families and patients
4. Folic Acid

Prevention

Avoiding ingestion of fava beans or oxidant substances