Glucose-6-phosphate dehydrogenase deficiency

Genetics

-Gene: G6PD (glucose-6-phosphate dehydrogenase; Xq28)

-XLR

Clinical findings/Dysmorphic features

-Hemolytic anemia (red blood cells destroyed faster than they get replaced) --> paleness, jaundice, dark urine, fatigue, shortness of breath, rapid heart rate

-Significant cause of mild to severe jaundice in newborns

-Many people never experience signs/symptoms and are unaware that they have the condition

-Some carrier females with symptoms

Etiology

-Most common enzymopathy; 400 million people worldwide affected; most frequent in parts of Africa, Asia, Mediterranean, Middle East; affects ~ 1 in 10 African American males in US

Pathogenesis

-Impaired ability of erythrocytes to form NADPH --> not enough to regenerate glutathione (natural antioxidant) --> toxic ROS accumulate --> hemolysis

Genetic testing/diagnosis

-G6PD enzyme activity level below 5 units per gram of hemoglobin constitutes deficiency

Others

-Triggers:

1) Food: Fava beans/inhaled pollen with high amounts of chemicals that are highly oxidative

2) Infections: immune system incites inflammatory response that generates oxidative species

3) Specific drugs: antibiotics and malaria medications