

Glucose-6-phosphate dehydrogenase deficiency

Glucose-6-phosphate dehydrogenase (G-6-PD) deficiency is a hereditary condition in which red blood cells break down when the body is exposed to certain drugs or the stress of infection. Babies with G6PD deficiency can have red blood cell break down due to stress of delivery.

Causes

G6PD deficiency occurs when a person is missing or doesn't have enough of an enzyme called glucose-6-phosphate dehydrogenase, which helps red blood cells work properly.

Too little G6PD leads to the destruction of red blood cells. This process is called hemolysis. When this process is actively occurring, it is called a hemolytic episode. The episodes are usually brief, because the body continues to produce new red blood cells, which have normal activity.

Red blood cell destruction can be triggered by infections, severe stress, certain foods, and certain drugs, including:

- Antimalarial drugs
- Aspirin
- Nitrofurantoin
- Nonsteroidal anti-inflammatory drugs (NSAIDs)
- Quinidine
- Quinine
- Sulfa drugs
- Nalidixic acid
- Dapsone
- Fava (Broad) beans
- Chinese herbal medicines
- Moth balls (Naphthaline)

In the United States, G6PD deficiency is more common among blacks than whites. Men are more likely to have this disorder than women.

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Symptoms

People with this condition do not display any signs of the disease until their red blood cells are exposed to certain chemicals in food or medicine, or to stress.

Symptoms include dark urine, enlarged spleen, fatigue, pallor, rapid heart rate, shortness of breath, and yellow skin color (jaundice). Babies with G6PD deficiency may have jaundice and anemia.

Exams and Tests

A blood test can be done to check the level of G6PD. Other tests that may be done include bilirubin level, complete blood count, urine tests, haptoglobin level, LDH test, methemoglobin reduction test, and reticulocyte count.

Treatment

Treatment may involve:

- Medicines to treat an infection, if present
- Stopping any drugs that are causing red blood cell destruction
- Transfusions, in some cases

Treatment for newborns may include a special light (phototherapy) if your baby has jaundice.

When to Contact a Your Doctor

Call for an appointment with your health care provider if your baby has any symptoms of this condition. Make sure you tell your baby's pediatrician and anyone caring for your baby that he/she has G6PD deficiency.

Prevention

Persons with G6PD deficiency must strictly avoid things that can trigger an episode. Talk to your health care provider about your medications.

Genetic counseling or testing may be available to those who have a family history of the condition.

Resources

Rady Children's Specialist: Division of Hematology
3010 Children's Way
San Diego, CA 92123
Office: (858) 966-5811

G6PD Deficiency Favism Association website: www.g6pd.org