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Glucose-6-phosphate dehydrogenase deficiency

Glucose-6-phosphate dehydrogenase (G6PD) deficiency is a condition in which red blood cells break down when the body is exposed to certain medicines or the stress of infection. It is hereditary, which means it is passed down in families.

Causes

G6PD deficiency occurs when a person is missing or does not have enough of an enzyme called glucose-6-phosphate dehydrogenase. This enzyme 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 most often brief. This is because the body continues to produce new red blood cells, which have normal G6PD activity.

Red blood cell destruction can be triggered by infections, certain foods (such as fava beans), and certain medicines, including:

- Antimalarial medicines such as quinine
- Aspirin (high doses)
- Nonsteroidal anti-inflammatory drugs (NSAIDs)
- Quinidine
- Sulfa medicines
- Antibiotics such as quinolones, nitrofurantoin

Other chemicals, such as those in mothballs, can also trigger an episode.

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

You are more likely to develop this condition if you:

- Are African American
- Are of Middle Eastern decent, particularly Kurdish or Sephardic Jewish
- Are male
- Have a family history of the deficiency

A form of this disorder is common in whites of Mediterranean descent. This form is also associated with acute episodes of hemolysis. Episodes are longer and more severe than in the other types of the disorder.

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.

Symptoms are more common in men and may include:

- Dark urine
- Fever
- Pain in abdomen
- Enlarged spleen and liver
- Fatigue
- Pallor
- Rapid heart rate
- Shortness of breath
- Yellow skin color (jaundice)

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 (CBC)
- Hemoglobin - urine
- Haptoglobin level
- Lactate dehydrogenase (LDH) test
- Methemoglobin reduction test
- Reticulocyte count

Treatment

Treatment may involve:

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

Outlook (Prognosis)

In most cases, hemolytic episodes go away on their own.

Possible Complications

In rare case, kidney failure or death may occur following a severe hemolytic event.

When to Contact a Medical Professional

Contact your health care provider if you have symptoms of this condition.

Contact your provider if you have been diagnosed with G6PD deficiency and symptoms do not disappear after treatment.

Prevention

People with G6PD deficiency must strictly avoid things that can trigger an episode. Talk to your provider about your medicines.

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

Alternative Names

G6PD deficiency; Hemolytic anemia due to G6PD deficiency; Anemia - hemolytic due to G6PD deficiency

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