G6PD Deficiency

KANAKIA Health Care

Dr. Swati Kanakia
MD, DCH, PhD
Pediatric Hematologist-Oncologist
w: www.kanakiahealthcare.com • t: 022 2417 3232 / 2410 1133

What is G6PD deficiency?

G6PD deficiency as the name suggests is a condition in which the body doesn't have enough glucose-6-phosphate dehydrogenase, or G6PD, which helps red blood cells (RBCs) function normally. This deficiency can cause premature breakdown of RBCs, which is called hemolysis. Hemolytic anemia occurs when RBCs are destroyed faster than the body can replace them, usually after exposure to certain medications, foods, or even infections.

What causes G6PD deficiency?

G6PD deficiency is a genetic condition that is passed down from either or both parents to their child. The defective gene that causes this deficiency is on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A striking characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.
Being a genetic disorder, there are more than 300 variants of this condition. The symptoms vary in their severity in different people according to the variant form of the genetic defect.

What are the symptoms of G6PD deficiency?

A child with G6PD deficiency is clinically normal most of the time. Certain triggers can lead to hemolysis of RBCs and symptoms. Some common triggers are:

- Bacterial and viral infections
- Severe stress
- Some drugs
  - Antimalarial drugs
    * Chloroquine
    * Primaquine
    * Quinine
  - Aspirin
  - Vitamin C
  - Nitrofurantoin
  - Nonsteroidal anti-inflammatory drugs (NSAIDs)
  - Antibiotics
    * Sulfa drugs
    * Chloramphenicol
    * Dapsone
    * Nalidixic acid
    * Ciprofloxacin
- Some chemicals e.g. naphthalene (found in mothballs)
- Some foods e.g. fava beans

During a hemolytic episode the symptoms may include:

- Pallor
- Extreme tiredness
- Dizziness
- Sudden rise in body temperature
- Rapid heartbeat
- Rapid breathing or shortness of breath
- Jaundice, or yellowing of the skin and eyes, particularly in newborns
- Backache

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- Pain in abdomen
- Enlarged spleen
- Dark, tea-colored urine

The symptoms usually go away in 3-6 weeks, once the trigger is removed or resolved.

How is G6PD deficiency diagnosed?

A simple blood test to check G6PD enzyme levels can diagnose G6PD deficiency. Other tests done are:
Complete blood count (CBC) – active G6PD shows presence of "Heinz bodies" (protein aggregates) within the RBCs
Hemoglobin – low
Reticulocyte count – as high as 30%
Liver function tests – unconjugated bilirubin is raised, but liver enzymes are normal, and to rule out other causes of liver damage
Haptaglobulin – decreased in hemolysis
Urine tests for free hemoglobin.

How is G6PD deficiency treated?

In a majority of case, all that is needed is to remove the trigger that caused the symptoms. To prevent hemolysis, the triggers must be avoided.
If an infection caused the trigger, then the infection has to be treated.
In cases of hemolytic anemia, more aggressive treatment may be required. This generally includes oxygen and blood transfusion.