



Glucose-6-phosphate dehydrogenase deficiency

Glucose-6-phosphate dehydrogenase deficiency is a genetic disorder that occurs almost exclusively in males. This condition mainly affects red blood cells, which carry oxygen from the lungs to tissues throughout the body. In affected individuals, a defect in an enzyme called glucose-6-phosphate dehydrogenase causes red blood cells to break down prematurely. This destruction of red blood cells is called hemolysis.

The most common medical problem associated with glucose-6-phosphate dehydrogenase deficiency is hemolytic anemia, which occurs when red blood cells are destroyed faster than the body can replace them. This type of anemia leads to paleness, yellowing of the skin and whites of the eyes (jaundice), dark urine, fatigue, shortness of breath, and a rapid heart rate. In people with glucose-6-phosphate dehydrogenase deficiency, hemolytic anemia is most often triggered by bacterial or viral infections or by certain drugs (such as some antibiotics and medications used to treat malaria). Hemolytic anemia can also occur after eating fava beans or inhaling pollen from fava plants (a reaction called favism).

Glucose-6-phosphate dehydrogenase deficiency is also a significant cause of mild to severe jaundice in newborns. Many people with this disorder, however, never experience any signs or symptoms and are unaware that they have the condition.

Frequency

An estimated 400 million people worldwide have glucose-6-phosphate dehydrogenase deficiency. This condition occurs most frequently in certain parts of Africa, Asia, the Mediterranean, and the Middle East. It affects about 1 in 10 African American males in the United States.

Causes

Glucose-6-phosphate dehydrogenase deficiency results from mutations in the *G6PD* gene. This gene provides instructions for making an enzyme called glucose-6-phosphate dehydrogenase. This enzyme is involved in the normal processing of carbohydrates. It also protects red blood cells from the effects of potentially harmful molecules called reactive oxygen species, which are byproducts of normal cellular functions. Chemical reactions involving glucose-6-phosphate dehydrogenase produce compounds that prevent reactive oxygen species from building up to toxic levels within red blood cells.

If mutations in the *G6PD* gene reduce the amount of glucose-6-phosphate dehydrogenase or alter its structure, this enzyme can no longer play its protective role. As a result, reactive oxygen species can accumulate and damage red blood cells. Factors such as infections, certain drugs, or ingesting fava beans can increase the

levels of reactive oxygen species, causing red blood cells to be destroyed faster than the body can replace them. A reduction in the number of red blood cells causes the signs and symptoms of hemolytic anemia.

Researchers believe that people who have a *G6PD* mutation may be partially protected against malaria, an infectious disease carried by a certain type of mosquito. A reduction in the amount of functional glucose-6-phosphate dehydrogenase appears to make it more difficult for this parasite to invade red blood cells. Glucose-6-phosphate dehydrogenase deficiency occurs most frequently in areas of the world where malaria is common.

Inheritance Pattern

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located 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 characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

Other Names for This Condition

- deficiency of glucose-6-phosphate dehydrogenase
- G6PD deficiency
- G6PDD
- glucose 6 phosphate dehydrogenase deficiency

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/geneticTesting](#)
- Genetic Testing Registry: Glucose 6 phosphate dehydrogenase deficiency
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0017758/>

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22glucose-6-phosphate+dehydrogenase+deficiency%22>

Other Diagnosis and Management Resources

- Baby's First Test
<https://www.babysfirsttest.org/newborn-screening/conditions/glucose-6-phosphate-dehydrogenase-deficiency>
- MedlinePlus Encyclopedia: Glucose-6-phosphate dehydrogenase deficiency
<https://medlineplus.gov/ency/article/000528.htm>
- MedlinePlus Encyclopedia: Glucose-6-phosphate dehydrogenase test
<https://medlineplus.gov/ency/article/003671.htm>
- MedlinePlus Encyclopedia: Hemolytic anemia
<https://medlineplus.gov/ency/article/000571.htm>
- MedlinePlus Encyclopedia: Newborn jaundice
<https://medlineplus.gov/ency/article/001559.htm>

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Glucose-6-phosphate dehydrogenase deficiency
<https://medlineplus.gov/ency/article/000528.htm>
- Encyclopedia: Glucose-6-phosphate dehydrogenase test
<https://medlineplus.gov/ency/article/003671.htm>
- Encyclopedia: Hemolytic anemia
<https://medlineplus.gov/ency/article/000571.htm>
- Encyclopedia: Newborn jaundice
<https://medlineplus.gov/ency/article/001559.htm>
- Health Topic: Anemia
<https://medlineplus.gov/anemia.html>
- Health Topic: G6PD Deficiency
<https://medlineplus.gov/g6pddeficiency.html>
- Health Topic: Newborn Screening
<https://medlineplus.gov/newbornscreening.html>

Genetic and Rare Diseases Information Center

- Glucose-6-phosphate dehydrogenase deficiency
<https://rarediseases.info.nih.gov/diseases/6520/glucose-6-phosphate-dehydrogenase-deficiency>

Educational Resources

- KidsHealth from the Nemours Foundation
<https://kidshealth.org/en/parents/g6pd.html>
- MalaCards: anemia, nonspherocytic hemolytic, due to g6pd deficiency
https://www.malacards.org/card/anemia_non_spherocytic_hemolytic_due_to_g6pd_deficiency
- Malaria Atlas Project
<https://map.ox.ac.uk/research-project/blood-disorders/>
- Orphanet: NON RARE IN EUROPE: Glucose-6-phosphate-dehydrogenase deficiency
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=362

Patient Support and Advocacy Resources

- Metabolic Support UK
<https://www.metabolicsupportuk.org/>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/glucose-6-phosphate-dehydrogenase-deficiency/>
- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/metaboli.html>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Glucosephosphate+Dehydrogenase+Deficiency%5BMAJR%5D%29+AND+%28glucose-6-phosphate+dehydrogenase+deficiency%5BTIAB%5D%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- GLUCOSE-6-PHOSPHATE DEHYDROGENASE
<http://omim.org/entry/305900>

Medical Genetics Database from MedGen

- Deficiency of glucose-6-phosphate dehydrogenase
<https://www.ncbi.nlm.nih.gov/medgen/473706>

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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/glucose-6-phosphate-dehydrogenase-deficiency>

Reviewed: May 2017

Published: September 10, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services