CONDITION

Glucose-6-phosphate dehydrogenase deficiency

An inherited enzyme deficiency that can make red blood cells break down after certain triggers.

WHAT IT IS
Inherited red blood cell enzyme deficiency

BODY SYSTEM
Blood and circulation

MAIN SYMPTOMS
Jaundice, dark urine, tiredness, breathlessness

WHAT CAUSES IT
Inherited change in the G6PD gene

HOW IT IS DIAGNOSED
Blood enzyme assay, sometimes genetic testing

MAIN TREATMENT
Trigger avoidance and supportive care during episodes

CAN IT BE CURED?
No - the inherited deficiency remains lifelong

IS IT AN EMERGENCY?
Sometimes - fainting, breathing trouble, or severe newborn jaundice

LONG-TERM OUTLOOK
Usually normal life with known trigger avoidance

Overview

Glucose-6-phosphate dehydrogenase deficiency, usually shortened to G6PD deficiency, is an inherited condition affecting red blood cells. G6PD is an enzyme that helps protect these cells from oxidative stress.

Oxidative stress can be produced by infections, certain medicines, fava beans, and some chemicals. If you have too little working G6PD, red blood cells can break down faster than your body can replace them. This is called hemolysis .

Rapid hemolysis can cause hemolytic anemia , meaning there are not enough red blood cells to carry oxygen around the body. It may also cause jaundice and dark urine.

Most people with G6PD deficiency feel well between episodes, and some never develop noticeable symptoms. Knowing about the condition can help you avoid triggers and receive appropriate treatment if hemolysis or newborn jaundice occurs.

Symptoms

Most older children and adults do not have ongoing symptoms. Symptoms usually appear after oxidative stress on the red blood cells.

During a hemolytic episode, symptoms may include:

Symptoms range from mild to severe. They often improve after the trigger has passed as the body makes new red blood cells. Medical assessment is still important because similar symptoms can have other causes.

In newborn babies, G6PD deficiency can contribute to jaundice during the first days of life. Bilirubin, a yellow substance produced when red blood cells break down, can sometimes rise to an unsafe level.

Yellowing can be harder to see on brown or black skin. Check the whites of the eyes and gums and notice changes from the baby's usual colour.

- Sudden tiredness, weakness, or paleness
- Yellow skin or yellowing in the whites of the eyes
- Dark brown, red, or tea-coloured urine
- Shortness of breath, a fast heartbeat, or dizziness
- Back or abdominal pain
- Fever or feeling unwell, especially if infection is the trigger

Causes and risk factors

G6PD deficiency is caused by a change in the G6PD gene , which provides instructions for making the enzyme.

The gene is on the X chromosome. People with one X chromosome are more likely to have clear deficiency if it carries the gene change. People with two X chromosomes may have no symptoms, mild deficiency, or significant deficiency because cells may use the two X chromosomes differently.

The condition is more common among people whose families come from parts of Africa, Asia,

the Mediterranean, and the Middle East, but it can occur in any background. Ancestry cannot confirm or rule it out.

Common triggers include:

Medicine risk varies by drug, dose, and degree of enzyme deficiency. Do not stop an essential prescribed medicine on your own. Ask a prescriber or pharmacist to check it against reliable G6PD-specific information.

- Infections, which are a frequent cause of episodes
- Fava beans, also called broad beans
- Certain medicines, including some antimalarial and antibacterial medicines
- Naphthalene, a chemical found in some mothballs

Diagnosis

G6PD deficiency is usually confirmed with a blood test that measures enzyme activity. A low result supports the diagnosis.

Timing matters. During or soon after hemolysis, many of the most affected red blood cells have already been destroyed. The younger cells left behind may have more enzyme activity, making the result appear normal. A recent blood transfusion can also affect the result.

If deficiency is still suspected after a normal test, testing may need to be repeated after the episode has settled and transfused cells no longer affect the sample.

Genetic testing can identify a change in the G6PD gene. It may help when an enzyme result is unclear, when testing relatives, or when a person with two X chromosomes has symptoms despite an uncertain result. It is not always required.

Blood tests during an episode may also check:

Newborn babies may need bilirubin measurements as well as assessment for G6PD deficiency.

- Red blood cell count and hemoglobin
- Bilirubin and other signs of red blood cell breakdown
- Kidney function
- Production of replacement red blood cells

Treatment and management

G6PD deficiency does not usually need daily treatment. Management focuses on avoiding triggers and treating hemolytic episodes.

If a medicine may be causing hemolysis, a healthcare professional will decide whether it should be stopped or changed. Infections should be assessed and treated because they can also trigger hemolysis.

A mild episode may settle with removal of the trigger, rest, fluids, and follow-up blood tests. More significant anemia may require hospital monitoring, oxygen, intravenous fluids, or a blood transfusion. Severe kidney problems are uncommon but require specialist care.

Newborn jaundice may be treated with phototherapy , which uses special light to help remove bilirubin. Very high levels may require intensive treatment, including an exchange transfusion in severe cases.

Tell prescribers, dentists, and pharmacists that you have G6PD deficiency. Keep the result in your medical record and consider carrying a medical alert card. Check every new prescription, over-the-counter medicine, herbal product, or supplement before taking it.

Medicine risk lists can conflict because evidence differs between drugs. Use a list recommended by a hematology, pharmacy, or genetics service rather than an unverified online list.

Complications

The main complication is acute hemolytic anemia. If many red blood cells break down quickly, the blood may not carry enough oxygen to the body's tissues.

Severe hemolysis can occasionally affect the kidneys as they filter products released from damaged red blood cells. Risk is greater with dehydration or extensive hemolysis.

Newborn jaundice is another important complication. If bilirubin becomes extremely high and is not treated promptly, it can enter the brain and cause permanent injury called kernicterus . Timely bilirubin checks and treatment greatly reduce this risk.

Most people do not have chronic anemia. Some uncommon G6PD gene changes cause ongoing red blood cell breakdown without an obvious trigger. This is called chronic nonspherocytic hemolytic anemia and usually needs specialist follow-up.

Prevention

G6PD deficiency cannot be prevented because it is inherited. It is not caused by anything a

parent did during pregnancy.

Episodes can often be prevented by avoiding known triggers. Do not eat fava beans if you have confirmed G6PD deficiency. Avoid direct exposure to naphthalene mothballs, especially around babies and children.

Tell healthcare professionals about the condition before receiving a new medicine. Some medicines, including certain antimalarial treatments, require G6PD testing before they can be used safely.

Infections cannot always be prevented, but seeking care when one develops may reduce the chance of severe illness. Routine vaccination can help prevent some infections.

If the condition runs in your family, genetic counselling can explain how it may be inherited. Testing may be considered for relatives, particularly before using a medicine known to cause hemolysis.

Living with it and what to expect

G6PD deficiency remains present for life, but the enzyme level does not usually worsen with age. Most people can take part in ordinary work, school, exercise, travel, and family life.

The main long-term task is learning which triggers apply to you. You do not need to avoid every medicine or food. Fava beans are the main food trigger, and unnecessary dietary restrictions may make life harder without adding protection.

After an episode, symptoms usually improve as the body replaces damaged red blood cells. Follow-up blood tests may be needed to confirm that anemia and kidney function have improved.

Routine specialist appointments are not always required if you remain well and do not have chronic anemia. Follow-up is more likely after a severe episode, uncertain test result, recurrent symptoms, or significant newborn jaundice.

Children should learn about the condition as they grow, including the need to check medicines and avoid fava beans and naphthalene. Schools and caregivers should know which symptoms to watch for.

When to get medical help

Arrange prompt medical advice if you develop new jaundice, dark urine, unusual paleness, or marked tiredness, especially after starting a medicine, eating fava beans, or having an

infection. Seek assessment even if a previous episode was mild.

A newborn with any jaundice should be assessed according to advice from the maternity or newborn care team. Seek urgent care if the yellow colour is increasing, spreading, or appearing during the first day of life.

Get emergency help for:

For routine care, discuss an unclear diagnosis, repeat testing, recurrent episodes, pregnancy planning, or medicine safety with an appropriate healthcare professional.

- Severe or rapidly worsening shortness of breath
- Fainting, confusion, chest pain, or extreme weakness
- Very little urine alongside dark urine or jaundice
- A baby who is difficult to wake, feeds poorly, arches their body, has unusual crying, or develops worsening jaundice

Common questions

Does G6PD deficiency go away?

No. It is caused by an inherited gene change and remains lifelong. Most people only have symptoms after exposure to a trigger.

What foods should I avoid with G6PD deficiency?

Fava beans, also called broad beans, are the established food trigger. Broad restrictions on other beans or foods are usually unnecessary unless a healthcare professional identifies another problem.

Which medicines are unsafe with G6PD deficiency?

Some antimalarial, antibacterial, and other medicines can cause hemolysis, but risk depends on the drug and sometimes the dose. Ask a pharmacist or prescriber to check each new medicine. A "sulfa" allergy and G6PD-related hemolysis are different issues.

Can women and girls have G6PD deficiency?

Yes. Although clear deficiency is more common in people with one X chromosome, women and girls can have low enzyme activity and clinically important hemolysis.

Is G6PD deficiency contagious?

No. It is inherited and cannot be caught from another person.

Can I pass G6PD deficiency to my children?

Yes. The chance and pattern depend on which parent carries the change and the child's chromosomes. Genetic counselling can explain the pattern for your family.

Sources

- MedlinePlus Genetics: Glucose-6-phosphate dehydrogenase deficiency , U.S. National Library of Medicine
- Luzzatto L, Ally M, Notaro R. Glucose-6-phosphate dehydrogenase deficiency , Blood , 2020;136(11):1225-1240
- Roper D, Layton M, Rees D, et al. Laboratory diagnosis of G6PD deficiency: a British Society for Haematology guideline , British Journal of Haematology , 2020;189(1):24-38
- Gammal RS, Pirmohamed M, Somogyi AA, et al. Expanded CPIC guideline for medication use in the context of G6PD genotype , Clinical Pharmacology & Therapeutics , 2023;113(5):973-985
- American Academy of Pediatrics. Clinical practice guideline revision: management of hyperbilirubinemia in newborn infants 35 or more weeks of gestation , Pediatrics , 2022;150(3):e2022058859