NEW YORK STATE | Department of Health | Wadsworth Center

Glucose 6-Phosphate Dehydrogenase Deficiency (G6PDD) Fact Sheet for Families

What is G6PDD?

Glucose 6-Phosphate Dehydrogenase Deficiency is also called G6PD deficiency or G6PDD. G6PDD is a genetic disease that can lead to low red blood cell counts. G6PDD causes red blood cells to be more easily damaged. Red blood cells are needed to carry oxygen through the body. If red blood cells are damaged, they break down (hemolysis) and the body has too few blood cells to carry oxygen to all parts of the body. This is called hemolytic anemia and can be mild or severe. Symptoms may include paleness, fast heart rate, dark urine, enlarged spleen, fever, tiredness, shortness of breath, and stomach pain.

Newborns with G6PDD are at risk for symptoms when red blood cells break down and a chemical called bilirubin gets released. High levels of bilirubin cause the skin and eyes to become yellow (jaundice). If bilirubin levels are too high, this can cause damage to the brain (kernicterus). Symptoms in babies may include poor feeding, vomiting, extreme sleepiness, and floppy muscles.

Many people with G6PDD don't have symptoms. In children and adults, it is rare to develop symptoms without a trigger, for example, certain foods, drugs, or infections. The most common triggers are fava beans and drugs for malaria. Symptoms usually last less than a week. In very rare cases, G6PDD may cause long-term symptoms without any known trigger.

Who does G6PDD affect?

G6PDD is very common and can affect anyone. Over four-hundred million people in the world have it. G6PDD is most common in families from:

- Africa
- The Mediterranean (examples: Greece, Italy)
- Asia
- The Middle East (examples: Syria, Turkey Iran, Saudi Arabia, Oman)

Who should be tested for G6PDD?

New York State law requires that all babies born in the state receive a "heel prick" soon after birth. This blood sample is used for newborn screening. This screen looks for many health disorders. The goal is to help find babies with treatable disorders. G6PDD is NOT screened for by the heel prick. In 2022 a new law in New York requires some babies to get extra testing for G6PDD.

Newborns should be tested for G6PDD if they have:

- Anemia because of red blood cell damage (hemolytic anemia)
- Yellowing of the skin/eyes (jaundice) because of red blood cell damage
- Jaundice that lasts more than one week after birth and is getting worse
- Been re-admitted to the hospital for jaundice
- A family history of G6PDD
- A risk for G6PDD based on ancestry (African, Mediterranean, Asian or Middle Eastern)

How can you test for G6PDD?

A blood sample can be used to test for G6PDD. This should be ordered by your baby's doctor. Some blood tests look at the enzyme level in the blood. Some blood tests look for genetic changes that can cause G6PDD (see next section). A test for G6PDD can be performed at any age.

What causes G6PDD?

Genetic diseases are caused by changes (variants) in a person's genetic instructions (DNA). A gene is a section of DNA that gives instructions to the body. For example, some genes are instructions to help the body break down food. G6PDD happens when a person has a variant in their *G6PD* gene that causes the gene to stop working. The *G6PD* gene is supposed to make an enzyme that protects red blood cells from damage. Without the enzyme, red blood cells are more easily damaged.

Both males and females can develop symptoms. Males have one copy of the *G6PD* gene, and females have two copies. Males with one *G6PD* gene variant have G6PDD and are at risk for symptoms. Males with G6PDD will pass their variant to all of their daughters and none of their sons. Females with one *G6PD* variant are called carriers. Some carriers have G6PDD and symptoms, and some carriers do not. It is rare for a female to have two *G6PD* variants. Females with two *G6PD* gene variants have G6PDD and are at risk for symptoms. Females can pass their variant(s) to both sons and daughters. Parents can pass G6PDD to their children even if the parents don't have symptoms.

How is G6PDD treated?

There is no cure for G6PDD because we can't change the genetic variants. However, there are treatments. Babies with G6PDD should be referred to a specialist. They may see a blood doctor (hematologist) or a genetic doctor (geneticist). A doctor will explain more about G6PDD. They may also order blood tests. G6PDD usually does not cause problems. The best treatment for G6PDD is to avoid triggers. Doctors may also prescribe folic acid or iron supplements. It is rare for a person with G6PDD to need a blood transfusion to treat severe anemia. Parents should talk with their child's doctor to determine the next steps. A doctor will determine if and what treatment is needed.

Resources for more information about G6PDD

Support groups can help connect families affected by G6PDD. Communities can offer support and the chance to talk to people with G6PDD experience. These groups offer resources to affected people, families, health care providers, and advocates:

G6PD Deficiency Foundation: https://g6pddf.org/
Baby's First Test: https://www.babysfirsttest.org/newborn-screening/conditions/glucose-6-phosphate-dehydrogenase-deficiency
G6PD Newborn screening: https://newbornscreening.hrsa.gov/conditions/glucose-6-phosphate-dehydrogenase-deficiency
Genetic and Rare Diseases Information Center (GARD): https://rarediseases.info.nih.gov/diseases/6520/glucose-6-phosphate-dehydrogenase-deficiency/
MedlinePlus: https://medlineplus.gov/genetics/condition/glucose-6-phosphate-dehydrogenase-deficiency/
National Organization for Rare Disorders, Inc. (NORD): https://rarediseases.org/rare-diseases/glucose-6-phosphate-dehydrogenase-deficiency/
Parents of Infants and Children with Kernicterus (PICK): https://pic-k.org/