G6PD Deficiency

G6PD deficiency is an inherited disorder. Lack of the enzyme G6PD means that red blood cells can be damaged and destroyed when the person is exposed to some chemicals (including some medicines) or during some infections. This can lead to moderate or severe haemolytic anaemia ('anaemia' means low levels of red blood cells, and 'haemolytic' means destroyed red blood cells).

Red blood cells are continuously being made in the bone marrow. Blood cells that are lost due to G6PD deficiency are replaced over the few weeks following the damage.

What is G6PD?

- G6PD, or Glucose-6-phosphate dehydrogenase, is an enzyme that is made in red blood cells.
- G6PD helps the body turn carbohydrates into energy.
- G6PD also helps protect red blood cells from 'oxidative' stress. Oxidative chemicals are made when some medicines are taken, when the body is exposed to some chemicals, or when the immune system is working hard to fight an infection.

 Causes of G6PD deficiency

- G6PD deficiency is an inherited disorder. The gene for it is on the X chromosome.
  - Females have two X chromosomes. If a female has the abnormal gene for G6PD on one of her X chromosomes, the other X chromosome with the normal gene can make enough G6PD, and she will not have the problem. If the female has two abnormal genes, she will have a G6PD deficiency.
  - Males have only one X chromosome, so if they have the abnormal gene, they will have the disorder. Therefore G6PD deficiency is much more common in males than females.
- The abnormal gene is most common in people from Africa, Mediterranean region, and South-East Asia.

Triggers for haemolytic anaemia

- Medicines that can trigger damage to the red blood cells include anti-malaria drugs, antibiotics that contain sulphur, aspirin and some medicines used for arthritis.
- It can also be triggered by naphthalene (in moth balls) and fava beans (sometimes G6PD deficiency is called 'favism').

Effects of G6PD deficiency
• Between episodes of anaemia, the person is completely healthy. In fact, having G6PD deficiency gives some protection against malaria.

• **The signs of haemolytic anaemia include:**
  - fatigue (tiredness)
  - paleness (in dark-skinned people, this may be most easily seen inside the mouth, on the tongue or lips)
  - shortness of breath
  - rapid heart rate
  - jaundice (yellowing of the skin and eyes)
  - dark urine.

• **If a person has an episode of haemolytic anaemia**
  - the trigger needs to be removed (eg. the medicine stopped or the infection treated).
  - sometimes a transfusion of red blood cells is needed.

**Further reading**

KidsHealth 'G6PD deficiency'
[http://kidshealth.org/PageManager.jsp?dn=KidsHealth&lic=1&ps=107&cat_id=130&article_set=20828](http://kidshealth.org/PageManager.jsp?dn=KidsHealth&lic=1&ps=107&cat_id=130&article_set=20828)

MedlinePlus Medical Encyclopedia 'Glucose 6 Phosphate-dehydrogenase deficiency'

*The information on this site should not be used as an alternative to professional care. If you have a particular problem, see a doctor, or ring the Parent Helpline on 1300 364 100 (local call cost from anywhere in South Australia).*