

Glucose-6-phosphate dehydrogenase (G6PD) deficiency

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is a common X-linked, hereditary defect in the G6PD gene. The G6PD enzyme is responsible for NADPH in red blood cells, which plays an important role in protecting the cells from oxidative damage. G6PD deficiency can accelerate the destruction of red blood cells and lead to haemolytic anaemia. It is a heterogeneous disorder with over 400 mutations. Enzyme-level deficiencies and clinical symptoms may vary in severity depending on the mutation and the individual patient.

Clinical manifestations

- Neonatal jaundice
- Drug-induced haemolysis
- Chronic nonspherocytic haemolytic anaemia
- Haemolytic anaemia associated with infection
- Favism (anaemia after ingesting fava beans).

Laboratory testing

Testing is not recommended following or during a haemolytic crisis, as enzyme levels may be falsely elevated due to a high number of reticulocytes. Enzyme activity may also increase when white cell and platelet counts are markedly high.

Biochemical testing can not reliably identify G6PD deficiency in heterozygous women, as enzyme activity may range from partially deficient to normal due to red cell mosaicism arising from random X-chromosome inactivation.

Treatment

Managing G6PD deficiency involves avoiding known oxidative stressors—including certain therapeutic agents—that may lead to haemolytic crisis.

These substances may include

- Drugs: Primaquine, sulphonamides, nitrofurans
- Chemicals: Naphthalene (moth balls)
- Foods: Fava beans
- Dyes: Henna

An extensive list of drugs and foodstuffs and their associated risks for G6PD-deficient people can be found at the G6PD Deficiency Association Web site: <http://www.g6pd.org>.