GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY AND ITS CLINICAL SIGNIFICANCE

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Glucose-6-phosphate dehydrogenase deficiency (G6PD deficiency) also known as favism (after the fava bean) is an X-linked recessive genetic condition that predisposes to hemolysis (spontaneous destruction of red blood cells) and resultant jaundice in response to a number of triggers, such as certain foods, illness, or medication. It is particularly common in people of Mediterranean and African origin. The condition is characterized by abnormally low levels of glucose-6-phosphate dehydrogenase, an enzyme involved in the pentose phosphate pathway that is especially important in the red blood cell. G6PD deficiency is the most common human enzyme defect. There is no specific treatment, other than avoiding known triggers.

Carriers of the G6PD allele appear to be protected to some extent against malaria, and in some cases dominant males have shown complete immunity to the disease. This accounts for the persistence of the allele in certain populations in that it confers a selective advantage. Most individuals with G6PD deficiency are asymptomatic. Symptomatic patients are almost exclusively male, due to the X-linked pattern of inheritance, but female carriers can be clinically affected due to unfavorable lyonization, where random inactivation of an X-chromosome in certain cells creates a population of G6PD-deficient red blood cells coexisting with normal red cells. A typical female with one affected X chromosome will show the deficiency in approximately half of her red blood cells. However, in rare cases, including double X deficiency, the ratio can be much more than half, making the individual almost as sensitive as a male.

Many substances are potentially harmful to people with G6PD deficiency. Antimalarial drugs that can cause acute hemolysis in people with G6PD deficiency includeprimaquine, pamaquine, and chloroquine. There is evidence that other antimalarials may also exacerbate G6PD deficiency, but only at higher doses. Sulfonamides (such as sulfanilamide, sulfamethoxazole, and mafenide), thiazolesulfone, methylene blue, and naphthalene should also be avoided by people with G6PD deficiency as they antagonize folate synthesis, as should certain analgesics (such as aspirin, phenazopyridine, and acetanilide) and a few non-sulfa antibiotics (nalidixic acid, nitrofurantoin, isoniazid, dapsone, and furazolidone). Henna has been known to cause haemolytic crisis in G6PD-deficient infants.