

one of these drugs, which generate hydrogen peroxide, in newborns with kidney injury and in adults with tumor lysis syndrome.

Although drug-induced AHA has been prominent in the study of G6PD deficiency, the most common clinical manifestations are in fact NNJ and favism, both of which are of public health importance in many populations. Contrary to beliefs that are still widespread, fava bean pollen inhalation does not cause favism, and other beans are safe.

A very small minority of subjects with G6PD deficiency have CNSHA of variable severity. The patient is nearly always a male, usually with a history of NNJ, who may present with anemia, unexplained jaundice, or gallstones later in life. The spleen may be enlarged. The severity of anemia ranges in different patients from borderline to transfusion dependent. The anemia is usually normo-macrocytic, with reticulocytosis. Bilirubin and LDH are increased. Although hemolysis is, by definition, chronic in these patients, they are also vulnerable to acute oxidative damage, and therefore the same agents that can cause AHA in people with the ordinary type of G6PD deficiency will cause severe exacerbations in people with CNSHA associated with G6PD deficiency. In some cases of CNSHA, the deficiency of G6PD is so severe in granulocytes that it limits their capacity to produce an oxidative burst, with consequent increased susceptibility to some bacterial infections.

Laboratory Diagnosis The suspicion of G6PD deficiency can be confirmed by semiquantitative methods often referred to as screening tests, which are suitable for population studies and can correctly classify male subjects, in the steady state, as G6PD normal or G6PD deficient. However, in clinical practice, a diagnostic test is usually needed when the patient has had a hemolytic attack: whereby the oldest, most G6PD-deficient red cells have been selectively destroyed, and young red cells, having higher G6PD activity, are being released into the circulation. Under these conditions, only a quantitative test can give a definitive result. In males, this test will identify normal hemizygotes and G6PD-deficient hemizygotes; among females, some heterozygotes will be missed, but those who are at most risk of hemolysis will be identified. Of course, G6PD deficiency also can be diagnosed by DNA testing.