Opis choroby *

Definicja

A rare constitutional hemolytic anemia due to an enzyme disorder characterized by severe glucose-6-phosphate dehydrogenase deficiency (typically <10% residual enzyme activity) associated with chronic non-spherocytic hemolytic anemia of highly variable severity. Patients are at risk of developing neonatal jaundice (potentially leading to kernicterus), gallstones, and reticulocytosis and splenomegaly. They have an increased susceptibility to oxidizing agents provoking episodes of acute hemolysis. Favism, which describes the occurrence of an acute hemolytic reaction in response to the ingestion of fava beans, is more common in infants and young children.

Dane

Klasyfikacja
Choroba

Synonimy
Class I G6PD deficiency
Ciężka niedokrwistość hemolityczna spowodowana niedoborem G6PD
Niedobór G6PD klasy I
Severe hemolytic anemia due to G6PD deficiency

Kod ORPHA
466026

Kod OMIM
300908

Kod ICD10
D55.0

Kod ICD11
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*Źródło

orphanet