Opis choroby *

Definicja

A rare, genetic, hemoglobinopathy characterized by generally mild clinical phenotype, high fetal hemoglobin levels and mild microcytosis and hypochromia. In some cases, acute sickle cell disease manifestations were reported, namely acute chest syndrome and acute pain crisis. The genotype is characterized by the combination of an HbS and HbF allele; symptoms depend on the degree of HbF:HbS expressivity with patients with more than 35% pancellular HbF expression being asymptomatic. Symptomatic patients have heterocellular expression of HbF.

Dane

Klasyfikacja
Choroba

Synonimy
HPFH-sickle cell disease syndrome
HPFH - niedokrwistość sierpowata

Kod ORPHA
251380

Kod OMIM
305435

Kod ICD10
D57.2

Kod ICD11
3A51.3

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*Źródło

orphanet