Opis choroby *

Definicja

A rare, genetic hemoglobinopathy usually characterized by mild microcytic hemolysis and, very rarely, vaso-occlusive complications. Severe manifestations have been reported, including hematuria, splenic infarction, acute chest syndrome, acute episodes of pain and reversible bone marrow necrosis. The genotype is characterized by an HbS allele in combination with an HbE variant (beta26glu>lys); symptoms are due to the low allelic expression of HbE leading to HbS predominance (65+/-5%).

Dane

Klasyfikacja
Choroba

Synonimy
HbSE disease
Choroba HbSE

Kod ORPHA
251375

Kod OMIM
-

Kod ICD10
D57.2

Kod ICD11
3A51.3

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*Źródło

orphanet