Opis choroby *

Definicja

A rare, genetic hemoglobinopathy characterized by all the characteristics of sickle cell anemia (SCA). Clinical course is similar to SCA, including acute episodes of pain, splenic infarction and splenic sequestration crisis, vaso-occlusive crisis, acute chest syndrome, ischemic brain injury, osteomyelitis and avascular bone necrosis. The genotype is characterized by an HbS allele in combination with the HbD variant, beta121Glu>Gln.

Dane

Klasyfikacja
Choroba

Synonimy
HbSD disease
Choroba HbSD

Kod ORPHA
251370

Kod OMIM
-

Kod ICD10
D57.2

Kod ICD11
3A51.3

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*Źródło

orphanet