Opis choroby *

Definicja

A rare hemoglobinopathy characterized by the presence of hemoglobin variants with structural abnormalities in the globin portion of the molecule which lead to auto-oxidation of heme iron, resulting in methemoglobinemia. Patients present with cyanosis for which no treatment is necessary. Mode of inheritance is autosomal dominant.

Dane

Klasyfikacja
Choroba

Synonimy
M hemoglobinopathy
Choroba hemoglobiy M
Dziedziczna methemoglobinemia z powodu mutacji hemoglobiny
Hemoglobinopatia M

Kod ORPHA
330041

Kod OMIM
617973

Kod ICD10
D74.0

Kod ICD11
3A92

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*Źródło

orphanet