

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 hemoglobiny M

Dziedziczna methemoglobinemia z powodu mutacji hemoglobiny

Hemoglobinopatia M

Kod ORPHA

330041

Kod OMIM

617973

Kod ICD10

D74.0

Kod ICD11

3A92

*Źródło

orphanet