

Opis choroby *

Definicja

A rare beta-thalassemia associated with another hemoglobin anomaly characterized by the presence of the hemoglobin Lepore variant in association with beta-thalassemia. Clinical presentation is highly variable, depending on the type of beta-thalassemia, and ranges from severe hypochromic microcytic anemia and complete transfusion dependency to moderate, compensated anemia without a need for regular blood transfusions.

Dane

Klasyfikacja

Choroba

Synonimy

HbLepore-beta-thalassemia syndrome

HbLepore - beta-talasemia

Lepore - beta-talasemia

Lepore-beta-thalassemia syndrome

Kod ORPHA

330032

Kod OMIM

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Kod ICD10

D56.8

Kod ICD11

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*Źródło

orphanet