Opis choroby *

Definicja

A rare, genetic metabolic disorder due to pyruvate kinase deficiency characterized by a variable degree of chronic nonspherocytic hemolytic anemia resulting in a variable clinical manifestations ranging from fatal anemia at birth to a to a fully compensated hemolysis without apparent anemia.

Dane

Klasyfikacja
Choroba

Synonimy
Pyruvate kinase deficiency of erythrocytes
Niedobó kinazy pirogronowej erytrocytów

Kod ORPHA
766

Kod OMIM
266200

Kod ICD10
D55.2

Kod ICD11
3A10.Y

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*Źródło

orphanet