

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

*Źródło

orphanet