Opis choroby *

Definicja

Prolidase deficiency is an inherited disorder of peptide metabolism characterized by severe skin lesions, recurrent infections (involving mainly the skin and respiratory system), dysmorphic facial features, variable cognitive impairment, and splenomegaly.

Dane

Klasyfikacja
Choroba

Synonimy
Hyperimidodipeptiduria
Hiperimidodipeptiduria

Kod ORPHA
742

Kod OMIM
170100

Kod ICD10
E72.8

Kod ICD11
5C50.F0

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*Źródło

orphanet