Opis choroby *

Definicja

A rare, severe, genetic form of hypophosphatasia (HPP) characterized by infantile rickets without elevated serum alkaline phosphatase (ALP) activity and a wide range of clinical manifestations due to hypomineralization.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Infantile Rathbun disease
Dziecięca choroba Rathburna
Dziecięca fosfoetanoloaminuria
Infantile phosphoethanolaminuria

Kod ORPHA
247651

Kod OMIM
241500

Kod ICD10
E83.3

Kod ICD11
5C64.3

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*Źródło

orphanet