

## 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 fosfoetanolaminuria  
Infantile phosphoethanolaminuria

#### Kod ORPHA

247651

#### Kod OMIM

241500

#### Kod ICD10

E83.3

#### Kod ICD11

5C64.3

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#### \*Źródło

orphanet