Opis choroby *

Definicja

A rare, genetic metabolic disorder characterized by reduced activity of unfractionated serum alkaline phosphatase (ALP) and various symptoms from life-threatening, severely impaired mineralization at birth to musculo-skeletal pain in adulthood.

Dane

Klasyfikacja
Choroba

Synonimy
HPP
Choroba Rathburna
Fosfoetanololaminuria
HPP
Phosphoethanolaminuria
Rathbun disease

Kod ORPHA
436

Kod OMIM
241500, 241510, 146300

Kod ICD10
E83.3

Kod ICD11
5C64.3

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*Źródło

orphanet