

Opis choroby *

Definicja

A very rare form of hypophosphatasia characterized by prenatal skeletal manifestations (limb shortening and bowing) that slowly resolve spontaneously and later may develop into the moderate childhood or adult forms of the disease.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

Prenatal benign Rathbun disease
Prenatalna łagodna choroba Rathburna
Prenatalna łagodna fosfoetanolaminuria
Prenatal benign phosphoethanolaminuria

Kod ORPHA

247638

Kod OMIM

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Kod ICD10

E83.3

Kod ICD11

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*Źródło

orphanet