Opis choroby *

Definicja

A rare, genetic, primary bone dysplasia syndrome characterized by multiple epiphyseal dysplasia, severely delayed ossification (mainly of the epiphyses, pubic symphysis, hands and feet), abnormal modeling of the bones in hands and feet, abnormal pelvis cartilage persistence, and mild growth retardation. Calcium, phosphate and vitamin D serum levels are typically within normal range, while parathyroid hormone serum levels are normal to slighly elevated. Oligodontia has been rarely associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
79106

Kod OMIM
600002

Kod ICD10
M85.8

Kod ICD11
FB83.0Y

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*Źródło

orphanet