

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 slightly 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

*Źródło

orphanet