Opis choroby *

Definicja

A rare bone development disorder characterized by mostly anterolateral bowing of the tibia usually evident at birth, with subsequent non-healing fractures and formation of a false joint (pseudoarthrosis), and instability and angulation at the pseudoarthrosis site. In the vast majority of patients the defect is unilateral, and more than half of the cases are associated with neurofibromatosis type 1.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Congenital pseudarthrosis of the tibia
Congenital pseudarthrosis of the tibia

Kod ORPHA
295018

Kod OMIM
-

Kod ICD10
Q74.2

Kod ICD11
LB9Y

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*Źródło

orphanet