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 Synonimy

Podtyp kliniczny Congenital pseudarthrosis of the tibia

Congenital pseudarthrosis of the tibia

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 295018
 Q74.2

Kod ICD11 LB9Y

*Źródło

orphanet