

Opis choroby *

Definicja

Severe lateral tibial bowing with short stature is a rare, genetic, primary bent bone dysplasia characterized by significant, uni-/bilateral, lateral tibial bowing localized to the distal two-thirds of the tibia, with respective cortical thickening and thinning of the inner and outer tibial curve, loss of normal trabecular bone, bilateral abnormalities of the tibial epiphyses and growth plates, as well as foot abnormalities, including abnormally high arches. Affected individuals have short stature with absence of other skeletal abnormalities.

Dane

Klasyfikacja

Choroba

Synonimy

Ciężkie boczne wygięcie kości piszczelowej-niski
wzrost-łagodnie odstająca łopaska-łagodna
dysmorfia twarzy

Kod ORPHA

324307

Kod OMIM

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Kod ICD10

Q68.4

Kod ICD11

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*Źródło

orphanet