Opis choroby *

Definicja

A rare primary bone dysplasia characterized by severe mesomelic shortness particularly of the lower limbs with distinctive triangular or rhomboid-shaped tibiae and fibulae, accompanied by bony protuberances and skin dimples. Additional manifestations include radioulnar synostosis, dislocation of the radial head, abnormalities of the hands (such as oligosyndactyly or fusiform-shaped fingers) and feet (pes equinovarus, synostoses of tarsals/metatarsals and phalanges), and dysmorphic facial features.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mesomelic dwarfism, Nievergelt type
Zespół Nievergelta
Nievergelt syndrome

Kod ORPHA
2633

Kod OMIM
163400

Kod ICD10
Q78.8

Kod ICD11
LD24.A

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*Źródło

orphanet