Opis choroby *

Definicja

A rare autosomal recessive acromesomelic dysplasia characterized by severe dwarfism at birth, abnormalities confined to limbs, severe shortening and deformity of long bones, fusion or absence of carpal and tarsal bones, ball shaped fingers and, occasionally, polydactyly and absent joints. As seen in acromesomelic dysplasia, Hunter-Thomson type and acromesomelic dysplasia, Maroteaux Type, facial features and intelligence are normal.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Chondrodysplasia, Grebe type
Chondrodysplazja, typu Grebe

Kod ORPHA
2098

Kod OMIM
200700

Kod ICD10
Q78.8

Kod ICD11
LD24.9

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*Źródło

orphanet