Opis choroby *

Definicja

A rare autosomal recessive acromesomelic dysplasia characterized by severe dwarfism (adult height approximately 120 cm) with abnormalities limited to the limbs (affecting the lower limbs more than upper limbs, with middle and distal segments being the most affected), severe shortening, absence or fusion of tubular bones of hands and feet and large joint dislocations. As seen in acromesomelic dysplasia, Grebe type and acromesomelic dysplasia, Maroteaux type, facial features and intelligence are normal.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Acromesomelic dwarfism
Karłowatość akromezomeliczna

Kod ORPHA
968

Kod OMIM
201250

Kod ICD10
Q78.8

Kod ICD11
LD24.9

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*Źródło

orphanet