Opis choroby *

Definicja

A rare autosomal recessive acromesomelic dysplasia characterized by severe dwarfism (adult height <120 cm), both axial and appendicular involvement (shortening of the middle and distal segments of limbs and vertebral shortening), and with normal facial appearance and intelligence. It is a less severe form than acromesomelic dysplasia, Grebe type and acromesomelic dysplasia, Hunter-Thomson type .

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
40

Kod OMIM
602875

Kod ICD10
Q77.8

Kod ICD11
LD24.9

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*Źródło

orphanet