

## 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