

## **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 radio-ulnar 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 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](#)

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