

## Opis choroby \*

### Definicja

A rare primary bone dysplasia due to matrilin-3 variants and characterized by disproportionate early-onset dwarfism, bowing of the lower limbs, short, wide and stocky long bones with severe epiphyseal and metaphyseal changes, lumbar lordosis, hypoplastic iliac bones, flat ovoid vertebral bodies and normal hands.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

SEMD, MATN3-related  
SEMD, typ matryliny-3  
SEMD, związana z MATN3  
SEMD, matrilin-3 type

#### Kod ORPHA

156728

#### Kod OMIM

608728

#### Kod ICD10

Q77.7

#### Kod ICD11

LD24.3

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#### \*Źródło

orphanet