

## Opis choroby \*

### Definicja

Spondyloepimetaphyseal dysplasia, Missouri type is characterized by moderate-to-severe metaphyseal changes, mild epiphyseal involvement, rhizomelic shortening of the lower limbs with bowing of the femora and/or tibiae, coxa vara, genu varum and pear-shaped vertebrae in childhood.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

SEMD type 2

SEMD typu 2

SEMD, typ Missouri

Spondyloepimetaphyseal dysplasia type 2

SEMD, Missouri type

Spondyloepimetaphyseal dysplasia type 2

#### Kod ORPHA

93356

#### Kod OMIM

602111

#### Kod ICD10

Q77.7

#### Kod ICD11

LD24.3

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#### \*Źródło

orphanet