

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

*Źródło

orphanet