Opis choroby *

Definicja

A rare spondyloepimetaphyseal dysplasia characterized by severe short-limb short stature beginning prenatally, joint hypermobility, dental abnormalities, dysmorphic facial features (including hypertelorism, midface hypoplasia, macroglossia, and prognathism), and other skeletal anomalies (such as atlantoaxial subluxation causing compression of the spinal cord, kyphoscoliosis, hip dislocation, or rocker-bottom feet). Mild intellectual disability may also be present.

Dane

Klasyfikacja
Choroba

Synonimy
Spondyloepimetaphyseal dysplasia, Menger type
Spondyloepimetaphyseal dysplasia, anauxetic type
Spondyloepimetaphyseal dysplasia, Menger type
Spondyloepimetaphyseal dysplasia, anauxetic type

Kod ORPHA
93347

Kod OMIM
617396

Kod ICD10
Q77.7

Kod ICD11
LD24.3

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*Źródło

orphanet