Opis choroby *

Definicja

A rare primary bone dysplasia characterized by short stature, joint laxity, vertebral anomalies, severe progressive spinal malalignment leading to spinal cord compression, progressive kyphoscoliosis, thoracic asymmetry, and elbow and foot deformities. Additional features include mild skin hyperelasticity, spatulate terminal phalanges, cleft palate and lip, structural cardiac malformations, and mild facial dysmorphism (oval face, prominent eyes with blue sclerae, and a long upper lip).

Dane

Klasyfikacja
Choroba

Synonimy
SEMD-JL
Dysplazja kręgowo-nasadowo-przynasadowa z wiotkością stawów typu 1
SEMDJL
SEMDJL1
SEMDJL1
Spondyloepimetaphyseal dysplasia with joint laxity type 1
Spondyloepimetaphyseal dysplasia with joint laxity, Beighton type

Kod ORPHA
93359

Kod OMIM
271640

Kod ICD10
Q77.7

Kod ICD11
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*Źródło

orphanet