

## 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	Synonimy
Choroba	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	Kod OMIM	Kod ICD10
93359	271640	Q77.7

### Kod ICD11

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### \*Źródło

orphanet