Opis choroby *

Definicja

A rare connective tissue disorder for which three subtypes exist, either related to the gene B4GALT7, B3GALT6 or SLC39A13, and for which the clinically overlapping characteristics include short stature (progressive in childhood), small joint hypermobility, skin hyperextensibility with soft, doughy skin especially on the hands and feet muscular hypotonia (ranging from congenitally severe to mild with later_onset), skeletal anomalies and, more variably, osteopenia, delayed motor development and bowing of the limbs. Gene-specific features, with variable presentation, are additionally observed in each subtype.

Dane

Klasyfikacja
Choroba

Synonimy
Spondylodysplastic EDS
spEDS
Spondylodysplastic EDS
spEDS

Kod ORPHA
536471

Kod OMIM
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Kod ICD10
Q79.6

Kod ICD11
LD28.1Y

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*Źródło

orphanet