Opis choroby *

Definicja

A rare systemic disease characterized by generalized joint hypermobility with recurrent joint dislocations, redundant and hyperextensible skin with poor wound healing and abnormal scarring, easy bruising, and osteopenia/osteoporosis. Additional manifestations include hypotonia, delayed motor development, foot deformities, prominent superficial veins in the chest region, vascular complications (like mitral valve prolapse and aortic root dilation), hernias, dental anomalies, scoliosis, and facial dysmorphisms (like high palate, micrognathia, narrow palate). Mode of inheritance is autosomal recessive.

Dane

Klasyfikacja
Choroba

Synonimy
AEBP1-related EDS
AEBP1-related Ehlers-Danlos syndrome
Classical-like EDS type 2
clEDS type 2
AEBP1-related EDS
AEBP1-related Ehlers-Danlos syndrome
Classical-like EDS type 2
clEDS type 2

Kod ORPHA
536532

Kod OMIM
618000

Kod ICD10
Q79.6

Kod ICD11
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*Źródło

orphanet