

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
cEDS type 2
AEBP1-related EDS
AEBP1-related Ehlers-Danlos syndrome
Classical-like EDS type 2
cEDS type 2

Kod ORPHA

536532

Kod OMIM

618000

Kod ICD10

Q79.6

Kod ICD11

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*Źródło

orphanet