Opis choroby *

Definicja

A rare systemic disease for which two subtypes exist, either related to the gene PLOD1 or FKBP22, and for which the clinically overlapping characteristics include congenital muscle hypotonia, congenital or early-onset kyphoscoliosis (progressive or non-progressive), and generalized joint hypermobility with dislocations/subluxations (in particular of the shoulders, hips, and knees). Additional features which may occur in both subtypes are skin hyperextensibility, easy bruising of the skin, rupture/aneurysm of a medium-sized artery, osteopenia/osteoporosis, blue sclerae, umbilical or inguinal hernia, chest deformity, marfanoid habitus, talipes equinovarus, and refractive errors. Gene-specific features, with variable presentation, are additionally observed in each subtype.

Dane

Klasyfikacja
Choroba

Synonimy
EDS VI
Ehlers-Danlos syndrome type 6
Ehlers-Danlos syndrome type 6
Kyphoscoliotic EDS
kEDS

Kod ORPHA
536545

Kod OMIM
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Kod ICD10
Q79.6

Kod ICD11
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*Źródło

orphanet