

## 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