

## Opis choroby \*

### Definicja

A rare subtype of kyphoscoliotic Ehlers-Danlos syndrome characterized by 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 common features 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. Subtype-specific manifestations include skin fragility, atrophic scarring, scleral/ocular fragility/rupture, microcornea, and facial dysmorphism (like low-set ears, epicanthal folds, down-slanting palpebral fissures, high palate). Molecular testing is obligatory to confirm the diagnosis.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Cutis hyperelastica  
EDS VIA  
EDS, oczno-skoliotyczny  
EDS, typ kifoskoliotyczny  
Zespół Ehlersa i Danlosa typu 6A  
Zespół Ehlersa i Danlosa, typ oczno-skoliotyczny  
EDS VIA  
Ehlers-Danlos syndrome type 6A  
Kyphoscoliotic EDS due to lysyl hydroxylase 1 deficiency  
Lysyl hydroxylase-deficient EDS  
Ocular-scoliotic EDS  
kEDS-PLOD1

#### Kod ORPHA

1900

#### Kod OMIM

225400

#### Kod ICD10

Q79.6

#### Kod ICD11

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