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 dysmorphology (like low‐set ears, epicanthal folds, down‐slanting palpebral fissures, high palate). Molecular testing is obligatory to confirm the diagnosis.

Dane

Klasyfikacja	Synonimy
Podtyp kliniczny	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	**Kod OMIM**	**Kod ICD10**
1900	225400	Q79.6

Kod ICD11

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