Opis choroby *

Definicja

A rare systemic disease for which two subtypes exist, either related to the gene <i>PLOD1</i> or <i>FKBP22</i>, 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

Kod OMIM

Kod ICD10

Q79.6

Kod ICD11

536545

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<u>*Źródło</u>

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