Opis choroby *

Definicja

A heterogeneous group of diseases characterized by fragility of the soft connective tissues resulting in widespread skin, ligament, joint, blood vessel and/or internal organ manifestations. Clinical spectrum is highly variable, ranging from mild skin and joint hyperlaxity to severe physical disability and life-threatening vascular complications. Overlap with osteogenesis imperfecta may be observed resulting in an EDS/osteogenesis imperfecta overlap phenotype. Diseases in this group include classical Ehlers-Danlos syndrome (EDS), musculocontractural EDS, hypermobile EDS, vascular EDS, arthrochalasia EDS, dermatosparaxis EDS, periodontal EDS, X-linked EDS, brittle cornea syndrome, classical-like EDS type 1 and type 2, cardiac-valvular EDS, spondylodysplastic EDS, myopathic EDS, and kyphoscoliotic EDS.

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
EDS
EDS

Kod ORPHA
98249

Kod OMIM
-

Kod ICD10
Q79.6

Kod ICD11
LD28.1

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*Źródło

orphanet