

Opis choroby *

Definicja

A rare systemic disease characterized by the association of the features of Ehlers-Danlos syndrome with those of osteogenesis imperfecta. Predominant clinical manifestations include generalized joint hypermobility and dislocations, skin hyperextensibility and/or translucency, easy bruising, and invariable association with mild signs of osteogenesis imperfecta, including short stature, blue sclera, and osteopenia or fractures.

Dane

Klasyfikacja

Choroba

Synonimy

EDS/OI syndrome

Zespół EDS/OI

Kod ORPHA

230857

Kod OMIM

619120

Kod ICD10

Q79.6

Kod ICD11

LD28.1Y

*Źródło

orphanet