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

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*Źródło

orphanet