Opis choroby *

Definicja

A rare type of Ehlers-Danlos syndrome characterized by childhood or adolescence onset of severe, intractable periodontitis, lack of attached gingiva, and presence of pretibial plaques. Additional manifestations are easy bruising, hypermobility mainly of the distal joints, skin hyperextensibility and fragility, abnormal scarring, recurrent infections, hernias, marfanoid facial features, acrogeria, and prominent vasculature.

Dane

Klasyfikacja
Choroba

Synonimy
EDS VIII
Zespół Ehlersa i Danlosa typ okołozębowy
Ehlers-Danlos syndrome type 8
Ehlers-Danlos syndrome, periodontitis type
Periodontal EDS
pEDS

Kod ORPHA
75392

Kod OMIM
130080

Kod ICD10
Q79.6

Kod ICD11
LD28.1Y

---

*Źródło

orphanet