Opis choroby *

Definicja

A rare, hereditary connective tissue disease characterized by severe ocular manifestations due to extreme corneal thinning and fragility with rupture in the absence of significant trauma, often leading to irreversible blindness. Extraocular manifestations comprise deafness, developmental hip dysplasia, and joint hypermobility.

Dane

Klasyfikacja
Choroba

Synonimy
Ehlers-Danlos syndrome type 6B

Kod ORPHA
90354

Kod OMIM
614170

Kod ICD10
Q79.6

Kod ICD11
LD28.1Y

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*Źródło

orphanet