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 Synonimy

Choroba Ehlers-Danlos syndrome type 6B

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 90354
 614170
 Q79.6

Kod ICD11 LD28.1Y

*Źródło

orphanet