Opis choroby *

Definicja

A rare group of genetic connective tissue disorders characterized by ophthalmic, auditory, orofacial and articular manifestations. The two main clinical forms are clinically distinguished by the vitreous phenotype; stickler type 1 by a vestigial vitreous gel in the immediate retrolental space, bordered by a distinct folded membrane, and Stickler type 2 by sparse and irregularly thickened bundles of ﬁbers throughout the vitreous cavity.

Dane

Klasyfikacja
Choroba

Synonimy
Hereditary progressive arthroophthalmopathy
Dziedziczna postępująca artrooftalmopatia

Kod ORPHA
828

Kod OMIM
STL1 (gen COL2A1) 108300, STL2 (gen COL11A1) 604841, STL4 (gen COL9A1) 614134, non-ocular Stickler syndrome (gen COL11A2), STL dziedziczony AR (gen COL9A3), STL5 (gen COL9A2) #614284, STL 6 (gen COL9A3) 620022

Kod ICD10
Q87.0

Kod ICD11
LD2F.1Y

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*Źródło

orphanet