

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 retroretinal 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

*Źródło

orphanet