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

Kod ORPHA
828

Kod OMIM
614284

Kod ICD10
Q87.0

Kod ICD11
LD2F.1Y

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*Źródło

orphanet