Opis choroby *

Definicja

A rare developmental defect with connective tissue involvement characterized by joint hyperextensibility and multiple dislocations of large joints, severe myopia, and short stature. Other common features include retinal detachment, iris and chorioretinal coloboma, kyphoscoliosis and other spine deformities, pectus carinatum, talipes equinovarus, and progressive hearing loss.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
527450

Kod OMIM
617662

Kod ICD10
Q87.5

Kod ICD11
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*Źródło

orphanet