

## 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