Opis choroby *

Definicja

A rare type of Stickler syndrome characterized by moderate to severe sensorineural hearing loss, high myopia, retinal degeneration, vitreous anomalies, and epiphyseal dysplasia. Midface hypoplasia, cleft palate, as well as additional skeletal manifestations (such as platyspondyly, scoliosis, and tibial and femoral bowing at birth) have also been observed.

Dane

Klasyfikacja
Podtyp kliniczny

Kod ORPHA
250984

Kod OMIM
614284

Kod ICD10
Q87.5

Kod ICD11
LD2F.1Y

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*Źródło

orphanet