Opis choroby *

Definicja

A very rare congenital limb malformation syndrome characterized by absence deformity of one leg, progressive scoliosis, short stature, and congenital cataract associated with dysplasia of the optic nerve. No intellectual deficit has been reported. There have been no further descriptions in the literature since 1968.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2310

Kod OMIM
246000

Kod ICD10
Q87.2

Kod ICD11
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*Źródło

orphanet