

Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability disorder characterized by the association of nonprogressive spastic quadripareisis, retinitis pigmentosa, intellectual disability, and variable deafness. There have been no further descriptions in the literature since 1976.

Dane

Klasyfikacja	Synonimy	
Choroba	Spastic quadriplegia-retinitis pigmentosa-intellectual disability syndrome Kwadriplegia spastyczna -zwyrodnienie barwnikowe siatkówki - niepełnosprawność intelektualna	
Kod ORPHA	Kod OMIM	Kod ICD10
3011	270950	G82.4

Kod ICD11

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*Źródło

orphanet