

Opis choroby *

Definicja

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

Dane

Klasyfikacja

Choroba

Synonimy

Spastic quadriplegia-retinitis pigmentosa-
intellectual disability syndrome

Kwadriplegia spastyczna -zwyrodnienie
barwnikowe siatkówki - niepełnosprawność
intelektualna

Kod ORPHA

3011

Kod OMIM

270950

Kod ICD10

G82.4

Kod ICD11

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*Źródło

orphanet