

Opis choroby *

Definicja

A rare, hereditary, cerebellar ataxia disorder characterized by late-onset spinocerebellar ataxia, manifesting with slowly progressive gait disturbances, dysarthria, limb and truncal ataxia, and smooth-pursuit eye movement disturbance, associated with a history of psychomotor delay from childhood. Mild atrophy of the cerebellar vermis and hemispheres is observed on brain imaging.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal recessive spinocerebellar ataxia type

11

Autosomalna recesywna ataksja rdzeniowo-
mózdkowa-11

SCAR11

SCAR11

Kod ORPHA

284271

Kod OMIM

614229

Kod ICD10

G11.1

Kod ICD11

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*Źródło

orphanet