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-móżdżkowa-11
SCAR11
SCAR11

Kod ORPHA
284271

Kod OMIM
614229

Kod ICD10
G11.1

Kod ICD11
-

---

*Źródło

orphanet