

## Opis choroby \*

### Definicja

A rare, genetic, autosomal recessive cerebellar ataxia disease characterized by adulthood-onset of slowly progressive spinocerebellar ataxia, manifesting with gait and appendicular ataxia, dysarthria, ocular movement anomalies (e.g. horizontal, vertical, and/or downbeat nystagmus, hypermetric saccades), increased deep tendon reflexes and progressive cognitive decline. Additional variable features may include proximal leg muscle wasting and fasciculations, pes cavus, inspiratory stridor, epilepsy, retinal degeneration and cataracts. Brain imaging reveals marked cerebellar atrophy and electromyography shows evidence of lower motor neuron involvement.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Autosomal recessive spinocerebellar ataxia type

10

Autosomalna recesywna ataksja rdzeniowo-mózdkowa-10

SCAR10

SCAR10

#### Kod ORPHA

284289

#### Kod OMIM

613728

#### Kod ICD10

G11.2

#### Kod ICD11

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#### \*Źródło

orphanet