

## Opis choroby \*

### Definicja

A rare hereditary ataxia characterized by a progressive cerebellar ataxia associated with disruption of visual fixation by saccadic intrusions (overshooting horizontal saccades with macrosaccadic oscillations and increased velocity of larger saccades). It presents with progressive gait, trunk and limb ataxia with pyramidal tract signs (increased tendon reflexes and Babinski sign), myoclonic jerks, fasciculations, cerebellar dysarthria, sensorimotor axonal neuropathy with impaired joint position, vibration, temperature, pain sensations, pes cavus, and saccadic intrusions with characteristic overshooting horizontal saccades, macrosaccadic oscillations, and increased velocity of larger saccades, without other eye movement disturbances.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

SCAR4

SCAR4

SCASI

SCASI

#### Kod ORPHA

95434

#### Kod OMIM

607317

#### Kod ICD10

G11.1

#### Kod ICD11

8A03.1Y

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#### [\\*Źródło](#)

orphanet