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