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-móżdżkowa-10
SCAR10
SCAR10

Kod ORPHA
284289

Kod OMIM
613728

Kod ICD10
G11.2

Kod ICD11
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*Źródło

orphanet