Opis choroby *

Definicja

A rare, genetic, autosomal recessive cerebellar ataxia disease characterized by slowly progressive spinocerebellar ataxia developing during childhood, manifesting with gait and limb ataxia, postural tremor, dysarthria, sensory alterations (e.g. decreased vibration sense), eye movement anomalies (i.e. nystagmus, saccadic pursuit, oculomotor apraxia), upper and lower limb fasciculations, and hyperreflexia with Babinski signs. Brain imaging reveals cerebellar, pontine, vermian and medullar atrophy.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive spinocerebellar ataxia type 7
Autosomalna recesywna ataksja rdzeniowo-móżdżkowa-7
SCAR7
SCAR7

Kod ORPHA
284324

Kod OMIM
609270

Kod ICD10
G11.1

Kod ICD11
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*Źródło

orphanet