Opis choroby *

Definicja

Spinocerebellar ataxia with axonal neuropathy type 1 is a rare, genetic neurological disorder characterized by a late childhood onset of slowly progressive cerebellar ataxia. Initial manifestations include weakness and atrophy of distal limb muscles, areflexia and loss of pain, vibration and touch sensations in upper and lower extremities. Gaze nystagmus, cerebellar dysarthria, peripheral neuropathy, stepagge gait and pes cavus develop as disease progresses. Cerebellar atrophy (especially of the vermis) is present in all affected individuals. Additional reported manifestations include seizures, mild brain atrophy, mild hypercholesterolemia and borderline hypoalbuminemia.

Dane

Klasyfikacja
Choroba

Synonimy
SCAN1
SCAN1

Kod ORPHA
94124

Kod OMIM
607250

Kod ICD10
G60.2

Kod ICD11
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*Źródło

orphanet