

Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 74 is a rare, genetic, spastic paraplegia-optic atrophy-neuropathy-related (SPOAN-like) disorder characterized by childhood onset of mild to moderate spastic paraparesis which manifests with gait impairment that very slowly progresses into late adulthood, hyperactive patellar reflex and bilateral extensor plantar response, in association with optic atrophy and typical symptoms of peripheral neuropathy, including reduced or absent ankle reflexes, lower limb atrophy and distal sensory impairment. Reduced visual acuity and pes cavus are frequently reported.

Dane

Klasyfikacja

Choroba

Synonimy

SPG74

SPG74

Kod ORPHA

468661

Kod OMIM

616451

Kod ICD10

G11.4

Kod ICD11

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*Źródło

orphanet