Opis choroby *

Definicja

A group of rare, genetic, neurodegenerative diseases characterized by an infancy- to childhood-onset of progressive spastic paraplegia (with delayed motor milestones, gait disturbances, hyperreflexia and extensor plantar responses), optic atrophy (which may be accompanied by nystagmus and visual loss) and progressive peripheral neuropathy (with sensory impairment and distal muscle weakness/atrophy in upper and lower extremities). Additional signs may include foot deformities, spinal defects (scoliosis, kyphosis), joint contractures, exaggerated startle response, speech disorders, hyperhidrosis, extrapyramidal signs and intellectual disability. In very rare cases, a variant phenotype with less prominent or absent optic atrophy and/or neuropathy may be observed.

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
SPOAN and SPOAN-related disorder
SPOAN i zaburzenia związane z SPOAN

Kod ORPHA
431320

Kod OMIM
-

Kod ICD10
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Kod ICD11
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*Źródło

orphanet