Opis choroby *

Definicja

A rare axonal hereditary motor and sensory neuropathy disease characterized by progressive, peripheral, axonal sensorimotor neuropathy (of variable severity), affecting predominantly the distal lower limbs, associated with progressive, variably severe, optic atrophy, which frequently leads to visual loss. Patients typically present distal limb muscle weakness and atrophy, hypo/areflexia, foot deformities, poor visual acuity (often with a central scotoma), nystagmus, and reduced peripheral and nocturnal vision. Additional reported manifestations include sensorineural hearing loss, major joint contractures, anosmia, scoliosis/lumbar hyperlordosis, cognitive impairment and vocal cord paresis.

Dane

Klasyfikacja
Choroba

Synonimy
CMT6
Choroba Charcota, Mariego i Tootha typu 6
CMT6
Neuropatia obwodowa i zanik nerwu wzrokowego
Charcot-Marie-Tooth disease type 6
HMSN 6
HMSN VI
Hereditary motor and sensory neuropathy type VI
Peripheral neuropathy and optic atrophy

Kod ORPHA
90120

Kod OMIM
616505

Kod ICD10
G60.0

Kod ICD11
8C20.1

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*Źródło

orphanet