

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

*Źródło

orphanet