

Opis choroby *

Definicja

A subtype of Charcot-Marie-Tooth disease type 4 characterized by childhood onset of severe, progressive, demyelinating sensorimotor neuropathy manifesting with distal muscle weakness and atrophy of hands and feet, distal sensory impairment (vibration and pinprick) of lower limbs, lactic acidosis, areflexia and severely reduced motor nerve conduction velocities (25 m/s or less). Patients may also present kyphoscoliosis, nystagmus, hearing loss, cerebellar ataxia and/or brain MRI abnormalities (putaminal and periaqueductal lesions).

Dane

Klasyfikacja

Choroba

Synonimy

CMT4K

Choroba Charcota, Mariego i Tootha typu 4K

Ciężka demielinizacyjna choroba Charcota,

Mariego i Tootha związana z SURF1

CMT4 związana z SURF1

CMT4K

Charcot-Marie-Tooth disease type 4K

SURF1-related CMT4

SURF1-related severe demyelinating Charcot-Marie-Tooth disease

Kod ORPHA

391351

Kod OMIM

616684

Kod ICD10

G60.0

Kod ICD11

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*Źródło

orphanet