

Opis choroby *

Definicja

Severe early-onset axonal neuropathy due to MFN2 deficiency is a rare axonal hereditary motor and sensory neuropathy characterized by early onset (<10 years) progressive distal muscle weakness and wasting of the lower limbs and later, to a lesser extent the upper limbs resulting in foot and wrist drop, areflexia, skeletal deformities (kyphoscoliosis, pes cavus with flattening, joint contractures), mild sensory impairment with vibration sense reduced to a greater extent than pain, optic atrophy and hearing loss. Wheelchair dependence by adolescence is usual and respiratory impairment with diaphragmatic paralysis may develop.

Dane

Klasyfikacja

Choroba

Synonimy

AR-CMT2, Ouvrier type

AR-CMT2, typ Ouvriera

Autosomalna recesywna choroba Charcota,

Mariego i Tootha, typ Ouvriera

SEOAN z powodu niedoboru MFN2

Autosomal recessive Charcot-Marie-Tooth

disease, Ouvrier type

SEOAN due to MFN2 deficiency

Kod ORPHA

90118

Kod OMIM

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Kod ICD10

G60.0

Kod ICD11

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*Źródło

orphanet