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