

Opis choroby *

Definicja

Hereditary motor and sensory neuropathy, Okinawa type is a rare, genetic, axonal hereditary motor and sensory neuropathy characterized by the adult-onset of slowly progressive, symmetric, proximal dominant muscle weakness and atrophy, painful muscle cramps, fasciculations and distal sensory impairment, mostly (but not exclusively) in individuals (and their descendants) from the Okinawa region in Japan. Absent deep tendon reflexes, elevated creatine kinase levels and autosomal dominant inheritance are also characteristic.

Dane

Klasyfikacja	Synonimy
Choroba	HMSNP Dziedziczna neuropatia ruchowa i czuciowa, typ proksymalny HMSNP Hereditary motor and sensory neuropathy, proximal type

Kod ORPHA	Kod OMIM	Kod ICD10
90117	604484	G60.0

Kod ICD11
8C20.Y

*Źródło

orphanet