

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 descendents) from the Okinawa region in Japan. Absent deep tendon reflexes, elevated creatine kinase levels and autosomal dominant inheritance are also characteristic.

Dane

Klasyfikacja

Choroba

Synonimy

HMSNP

Dziedziczna neuropatia ruchowa i czuciowa, typ proksymalny

HMSNP

Hereditary motor and sensory neuropathy, proximal type

Kod ORPHA

90117

Kod OMIM

604484

Kod ICD10

G60.0

Kod ICD11

8C20.Y

[*Źródło](#)

orphanet