

Opis choroby *

Definicja

Charcot-Marie-Tooth disease-deafness-intellectual disability syndrome is a rare demyelinating hereditary motor and sensory neuropathy characterized by early-onset, slowly progressive, distal muscular weakness and atrophy with no sensory impairment, congenital sensorineural deafness and mild intellectual disability (with absence of normal speech development). The absence of large myelinated fibers on sural nerve biopsy is equally characteristic of the disease.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

CMT-deafness-intellectual disability syndrome

Dziedziczna neuropatia ruchowa i czuciowa z głuchotą, niepełnosprawnością intelektualną oraz brakiem dużych włókien mielinowych

Charcot-Marie-Tooth disease-hearing loss-intellectual disability syndrome

Hereditary motor and sensory neuropathy with deafness, intellectual disability and absent sensory large myelinated fibers

Hereditary motor and sensory neuropathy with hearing loss, intellectual disability and absent sensory large myelinated fibers

Kod ORPHA

90103

Kod OMIM

214370

Kod ICD10

G60.0

Kod ICD11

8C20.Y

*Źródło

orphanet