

Opis choroby *

Definicja

A rare subtype of CMT1 characterized by a variable clinical presentation. Onset within the first two years of life with a delay in walking is not uncommon; however, onset may occur later. CMT1E is caused by point mutations in the *PMP22* (17p12) gene. The disease severity depends on the particular *PMP22* mutation, with some cases being very mild and even resembling hereditary neuropathy with liability to pressure palsies, while others having an earlier onset with a more severe phenotype (reminiscent of Dejerine-Sottas syndrome) than that seen in CMT1A, caused by gene duplication. These severe cases may also report deafness and much slower motor nerve conduction velocities compared to CMT1A patients.

Dane

Klasyfikacja

Choroba

Synonimy

CMT1E

Choroba Charcota, Mariego i Tootha - głuchota

CMT1E

Charcot-Marie-Tooth disease-deafness
syndrome

Charcot-Marie-Tooth disease-hearing loss
syndrome

Kod ORPHA

90658

Kod OMIM

118300

Kod ICD10

G60.0

Kod ICD11

8C20.0

*Źródło

orphanet