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

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*Źródło

orphanet