Opis choroby *

Definicja

Charcot-Marie-Tooth disease type 4E (CMT4E) is a congenital, hypomyelinating subtype of Charcot-Marie-Tooth disease type 4 characterized by a Dejerine-Sottas syndrome-like phenotype (incl. hypotonia and/or delayed motor development in infancy), extremely slow nerve conduction velocities, potential respiratory dysfunction, cranial nerve involvement, and the typical CMT phenotype, i.e. distal muscle weakness and atrophy, sensory loss, and foot deformity.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive congenital hypomyelinating neuropathy
Autosomalna recesywna wrodzona neuropatia hipomielinizująca
CMT4E
CMT4E

Kod ORPHA
99951

Kod OMIM
605253

Kod ICD10
G60.0

Kod ICD11
8C20.0

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*Źródło

orphanet