

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

*Źródło

orphanet