

Opis choroby *

Definicja

Charcot-Marie-Tooth disease, type 2B2 (CMT2B2, also referred to as CMT4C3) is an axonal CMT peripheral sensorimotor polyneuropathy that has been described in a large consanguineous Costa Rican family of Spanish ancestry.

Dane

Klasyfikacja

Choroba

Synonimy

AR-CMT2B2

AR-CMT2B2

Autosomalna recesywna aksonalna choroba

Charcota, Mariego i Tootha typu 2B2

Autosomalna recesywna aksonalna CMT4C3

Autosomal recessive axonal CMT4C3

Autosomal recessive axonal Charcot-Marie-Tooth disease type 2B2

Kod ORPHA

101101

Kod OMIM

605589

Kod ICD10

G60.0

Kod ICD11

8C20.1

*Źródło

orphanet