

Choroba Charcota, Mariego i Tootha typu 4H

Kod Orpha: 99954 Kod OMIM: 609311

Opis choroby *

Definicja

Charcot-Marie-Tooth disease type 4H is a subtype of Charcot-Marie-Tooth disease type 4 characterized by onset before two years of age of severe, slowly progressive, demyelinating sensorimotor neuropathy manifesting with delayed motor development (walking), unsteady gait, distal muscle weakness and atrophy (more prominent in the lower limbs), areflexia, mild symmetrical stocking-distribution hypoesthesia, and skeletal malformations (incl. kyphoscoliosis, short neck, pes cavus and pes equinus). Severely reduced nerve conduction velocities are associated.

Dane

Klasyfikacja

Choroba

Synonimy

CMT4H

CMT4H

Kod ORPHA

99954

Kod OMIM

609311

Kod ICD10

G60.0

Kod ICD11

8C20.0

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.