

Opis choroby *

Definicja

A rare, genetic, subtype of autosomal dominant Charcot-Marie-Tooth disease type 2 characterized by early childhood-onset of slowly progressive, predominantly distal, lower limb muscle weakness and atrophy, delayed motor development, variable sensory loss, and pes cavus in the presence of normal or near-normal nerve conduction velocities. Additional variable features may include proximal muscle weakness, abnormal gait, arthrogryposis, scoliosis, cognitive impairment, and spasticity.

Dane

Klasyfikacja

Choroba

Synonimy

CMT2O

CMT2O

Kod ORPHA

284232

Kod OMIM

614228

Kod ICD10

G60.0

Kod ICD11

8C20.1

*Źródło

orphanet