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

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*Źródło

orphanet