Opis choroby *

Definicja

A subtype of autosomal recessive intermediate Charcot-Marie-Tooth (CMT) disease characterized by severe, early childhood-onset CMT neuropathy with prominent pes equinovarus deformity and impairment of hand muscles. Nerve conduction velocities usually range between 25-35 m/s and both axonal and demyelinating changes are observed on peripheral nerve pathology.

Dane

Klasyfikacja
Choroba

Synonimy
RI-CMT type A
RI-CMT typu A

Kod ORPHA
217055

Kod OMIM
608340

Kod ICD10
G60.0

Kod ICD11
8C20.2

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*Źródło

orphanet