

## 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

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#### Kod ORPHA

217055

#### Kod OMIM

608340

#### Kod ICD10

G60.0

#### Kod ICD11

8C20.2

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#### \*Źródło

orphanet