

## Opis choroby \*

### Definicja

Charcot-Marie-Tooth disease type 4B1 (CMT4B1) is a subtype of Charcot-Marie-Tooth disease type 4 characterized by an early childhood-onset of severe, demyelinating sensorimotor neuropathy, various degrees of complex myelin outfoldings seen on peripheral nerve biopsy, very slow, and often undetectable, nerve conduction velocities, and the typical CMT phenotype (i.e. distal muscle weakness and atrophy, sensory loss, and frequent pes cavus). Other reported features include facial weakness, vocal cord paresis, respiratory difficulties, and skeletal deformities (e.g. chest deformities, claw hands, pes equinovarus).

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

CMT4B1

CMT4B1

#### Kod ORPHA

99955

#### Kod OMIM

601382

#### Kod ICD10

G60.0

#### Kod ICD11

8C20.0

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#### \*Źródło

orphanet