Opis choroby *

Definicja

Autosomal recessive intermediate Charcot-Marie-Tooth disease type D is a rare hereditary motor and sensory neuropathy characterized by childhood onset of unsteady gait, pes cavus, frequent falls and foot dorsiflexor weakness slowly progressing to distal upper and lower limb muscle weakness and atrophy, distal sensory impairment and reduced tendon reflexes. Additional symptoms may include bilateral sensorineural hearing impairment and neuropathic pain.

Dane

Klasyfikacja
Choroba

Synonimy
RI-CMT type D
RI-CMT typu D

Kod ORPHA
435998

Kod OMIM
616039

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet