Opis choroby *

Definicja

A rare autosomal dominant hereditary axonal motor and sensory neuropathy characterized by childhood onset of slowly progressive distal muscle weakness and atrophy primarily affecting the lower limbs, associated with sensory impairment and ataxia presenting with an unsteady, broad-based gait and frequent falls. Additional signs include decreased deep tendon reflexes and hand tremor.

Dane

Klasyfikacja
Choroba

Synonimy
CMT2 due to DGAT2 mutation
CMT2 z powodu mutacji w DGAT2

Kod ORPHA
487814

Kod OMIM
-

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet