

Choroba Charcota, Mariego i Tootha typu 2S

Kod Orpha: 443073 Kod OMIM: 616155

Opis choroby *

Definicja

A rare subtype of axonal hereditary motor and sensory neuropathy characterized by progressive distal muscle weakness and atrophy of both the lower and upper limbs, absent or reduced deep tendon reflexes, mild sensory loss, foot drop, and pes cavus leading eventually to wheelchair dependence. Some patients present with early hypotonia and delayed motor development. Scoliosis and variable autonomic disturbances may be associated.

Dane

Klasyfikacja

Choroba

Synonimy

CMT2S

CMT2S

Kod ORPHA

443073

Kod OMIM

616155

Kod ICD10

G60.0

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.