Opis choroby *

Definicja

A rare autosomal recessive axonal hereditary motor and sensory neuropathy characterized by motor-predominant axonal polyneuropathy due to a defect in copper metabolism. Patients become symptomatic in infancy or childhood with subtle motor delay or regression, manifesting with progressive weakness, muscle wasting, and absent reflexes in the lower and upper extremities. In addition, vibratory sensation is mildly diminished. Involvement of the face with weakness and fasciculation of facial muscles has also been described.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive axonal CMT due to copper metabolism defect
Autosomal recessive axonal CMT due to copper metabolism defect

Kod ORPHA
521411

Kod OMIM
-

Kod ICD10
G60.0

Kod ICD11
-

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*Źródło

orphanet