Opis choroby *

Definicja

A rare genetic neuromuscular disease characterized by length-dependent axonal motor neuropathy predominantly affecting the lower limbs, in combination with a myopathy with morphological features of myofibrillar myopathy with aggregates and rimmed vacuoles. Age of onset is typically in the second to third decade of life. Patients present with slowly progressive muscle weakness and atrophy initially affecting the distal lower limbs and later progressing to involve proximal limbs and also truncal muscles. There is no involvement of respiratory and cardiac muscles.

Dane

Klasyfikacja
Choroba

Kod ORPHA
476093

Kod OMIM
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Kod ICD10
G60.8

Kod ICD11
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*Źródło

orphanet