

Opis choroby *

Definicja

Distal myopathy with anterior tibial onset is a rare, genetic neuromuscular disease characterized by a progressive muscle weakness starting in the anterior tibial muscles, later involving lower and upper limb muscles, associated with an increased serum creatine kinase levels and absence of dysferlin on muscle biopsy. Patients become wheelchair dependent.

Dane

Klasyfikacja

Choroba

Synonimy

Distal anterior compartment myopathy
Distal anterior compartment myopathy

Kod ORPHA

178400

Kod OMIM

606768

Kod ICD10

G71.0

Kod ICD11

8C75

*Źródło

orphanet