Opis choroby *

Definicja

A rare, genetic, non-dystrophic myofibrillar myopathy disorder characterized by late-adult onset of distal and/or proximal limb muscle weakness with initial involvement of posterior lower leg muscles, medial gastrocnemius and soleus. Patients present with ankle weakness followed by weakness of finger and wrist extensors and later on of proximal muscles. Ambulation is usually preserved. Late-onset associated cardiomyopathy and/or neuropathy has been reported in a minority of cases.

Dane

Klasyfikacja
Choroba

Synonimy
ZASP-related myofibrillar myopathy
Miopatia miofibrylarna związana z ZASP

Kod ORPHA
98912

Kod OMIM
609452

Kod ICD10
G71.8

Kod ICD11
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*Źródło

orphanet