

Opis choroby *

Definicja

Muscle filaminopathy is a rare myofibrillar myopathy characterized by slowly progressive, proximal skeletal muscle weakness, which is initially more prominent in lower extremities and involves upper extremities with disease progression. Patients present with difficulty climbing stairs, a waddling gait, marked winging of scapula, lower back pain, paresis of limb girdle musculature, hypo-/areflexia and/or mild facial muscle weakness in rare cases. Respiratory muscle weakness is common and cardiac anomalies (conduction blocks, tachycardia, diastolic dysfunction, left ventricular hypertrophy) have been reported in some cases.

Dane

Klasyfikacja

Choroba

Synonimy

Filaminopatia mięśniowa
FLNC-associated myofibrillar myopathy
Filamin C-related filaminopathy
MFM5

Kod ORPHA

171445

Kod OMIM

609524

Kod ICD10

G71.8

Kod ICD11

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*Źródło

orphanet