

Opis choroby *

Definicja

Spheroid body myopathy is a rare form of myofibrillar myopathy characterized by predominantly proximal muscle weakness (that could be either non- or slowly progressive), associated with spheroid body inclusions (composed of myofilamentous material within individual muscle fibers) in skeletal muscle biopsy. Presentation is varied and may range from asymptomatic to severe muscle weakness that manifests with absent Achilles reflexes, gait abnormality and/or other motor incapacitations.

Dane

Klasyfikacja

Choroba

Kod ORPHA

268129

Kod OMIM

182920

Kod ICD10

G71.8

Kod ICD11

8C72.0Y

*Źródło

orphanet