

Miopatia dystalna zależna od syntetazy adenylbursztynianowej-1

Kod Orpha: 482601 Kod OMIM: 617030

Opis choroby *

Definicja

A rare autosomal recessive distal myopathy characterized by slowly progressive diffuse muscle weakness in childhood, followed by predominantly distal muscle weakness in adolescence, and quadriceps muscle weakness in the fourth decade. Facial muscle weakness is commonly reported. Muscle biopsy shows fiber size variation, increased internal nuclei, fiber splitting, rimmed vacuoles, and focal endomysial fibrosis.

Dane

Klasyfikacja

Choroba

Synonimy

ADSSL1-related distal myopathy

Miopatia dystalna zależna od ADSSL1

Kod ORPHA

482601

Kod OMIM

617030

Kod ICD10

G71.0

Kod ICD11

-

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.