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 Synonimy

Choroba ADSSL1-related distal myopathy

Miopatia dystalna zależna od ADSSL1

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 482601
 617030
 G71.0

Kod ICD11

-

*Źródło

orphanet