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
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*Źródło

orphanet