Opis choroby *

Definicja

Myosclerosis is a rare, genetic, non-dystrophic myopathy characterized by early, diffuse, progressive muscle and joint contractures that result in severe limitation of movement of axial, proximal, and distal joints, walking difficulties in early childhood and toe walking. Patients typically present thin, sclerotic muscles with a woody consistency, mild girdle and proximal limb weakness with moderate distal weakness and scoliosis. Muscle biopsy shows partial collagen VI deficiency at the myofiber basement membrane and absent collagen VI around most endomysial/perimysial capillaries.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital myosclerosis, Löwenthal type
Wrodzone stwardnienie mięśni

Kod ORPHA
289380

Kod OMIM
255600

Kod ICD10
G71.8

Kod ICD11
8C72.1

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*Źródło

orphanet