## 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 Congenital myosclerosis, Löwenthal type

Wrodzone stwardnienie mięśni

Kod ORPHA 289380

**Kod OMIM** 255600

Synonimy

**Kod ICD10** G71.8

**Kod ICD11** 8C72.1

\*Źródło

orphanet