

# Stwardnienie mięśni

Kod Orpha: 289380 Kod OMIM: 255600

## 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

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.