Myosclerosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Myosclerosis. ORPHA:289380

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.