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# Distal anoctaminopathy

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base*. [Distal anoctaminopathy](#). ORPHA:399096

Distal anoctaminopathy is a rare, autosomal recessive distal myopathy characterized by early adult-onset, slowly progressive, often asymmetrical, lower limb muscle weakness initially affecting the calves (with relative anterior muscle sparing) and later proximal muscle involvement, as well as highly elevated creatine kinase (CK) serum levels.