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# Childhood-onset autosomal recessive myopathy with external ophthalmoplegia

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Childhood-onset autosomal recessive myopathy with external ophthalmoplegia. ORPHA:363677*

A rare, genetic, non-dystrophic myopathy disease characterized by childhood-onset severe external ophthalmoplegia, typically without ptosis, associated with mild, very slowly progressive muscular weakness and atrophy, involving the facial, neck flexor and limb (upper > lower, proximal > distal) muscles. Muscle biopsy shows type 1 fiber uniformity, absent, or abnormally small, type 2A fibers, increased variability of fiber size, internalized nuclei and/or fatty infiltration.