# Congenital myopathy with internal nuclei and atypical cores

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital myopathy with internal nuclei and atypical cores. ORPHA:319160*

Congenital myopathy with internal nuclei and atypical cores is a rare genetic skeletal muscle disease characterized by neonatal hypotonia, distal more than proximal muscle weakness, progressive exercise intolerance with prominent myalgias, and mild-to-moderate overall motor impairment with preserved ambulation. Face, extraocular, cardiac, and respiratory muscles are unaffected. Mild cognitive impairment is also noted in most patients.