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Cylindrical spirals myopathy

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Cylindrical spirals myopathy. ORPHA:171886*

Cylindrical spirals myopathy is a rare form of congenital myopathy characterized by global muscle weakness, hypotonia, myotonia and cramps in the presence of cylindrical, spiral-shaped inclusions (located in the central and/or subsarcolemmal areas of muscle fibers) in skeletal muscle biopsy. Abnormal gait, scoliosis, epileptic encephalopathy and psychomotor delay may be associated.