

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Kod ORPHA

171886

Kod OMIM

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Kod ICD10

G71.2

Kod ICD11

8C72.0Y

*Źródło

orphanet