

PubMed

**Format:** Abstract

Full text links

Neuropathol Appl Neurobiol. 2017 Feb;43(1):5-23. doi: 10.1111/nan.12369.

Myopathology in congenital myopathies.

Sewry CA^{1,2}, Wallgren-Pettersson C³.

Author information

Abstract

Congenital myopathies are clinically and genetically a heterogeneous group of early onset neuromuscular disorders, characterized by hypotonia and muscle weakness. Clinical severity and age of onset are variable. Many patients are severely affected at birth while others have a milder, moderately progressive or nonprogressive phenotype. Respiratory weakness is a major clinical aspect that requires regular monitoring. Causative mutations in several genes have been identified that are inherited in a dominant, recessive or X-linked manner, or arise de novo. Muscle biopsies show characteristic pathological features such as nemaline rods/bodies, cores, central nuclei or caps. Small type 1 fibres expressing slow myosin are a common feature and may sometimes be the only abnormality. Small cores (minicores) devoid of mitochondria and areas showing variable myofibrillar disruption occur in several neuromuscular disorders including several forms of congenital myopathy. Muscle biopsies can also show more than one structural defect. There is considerable clinical, pathological and genetic overlap with mutations in one gene resulting in more than one pathological feature, and the same pathological feature being associated with defects in more than one gene. Increasing application of whole exome sequencing is broadening the clinical and pathological spectra in congenital myopathies, but pathology still has a role in clarifying the pathogenicity of gene variants as well as directing molecular analysis.

KEYWORDS: central nuclei; congenital myopathies; cores; fibre-type disproportion; myotubular; nemalinePMID: 27976420 DOI: [10.1111/nan.12369](https://doi.org/10.1111/nan.12369)

[Indexed for MEDLINE]

Publication type, MeSH terms

LinkOut - more resources