An Unusual Pattern of Nemaline Myopathy

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Abstract

Nemaline myopathies are a heterogenous group of congenital myopathies caused by inherited mutations in at least twelve genes. A characteristic morphological feature of this group of diseases is the presence of nemaline rods within myofibres. The rods are derived from Z-line material and have a lattice substructure similar to Z-lines when viewed by electron microscopy. The case presented is a muscle biopsy from a ventilator-dependent 2-month-old infant with progressive generalised weakness. Light microscopy revealed severe myopathic features, and nemaline rods were identified using the modified Gomori trichrome staining technique. Electron microscopy revealed diseased myofibres characterised by abundant nemaline rods and swathes of filamentous aggregates (Figure 1). The unusual ultrastructure was suggestive of a nemaline myopathy caused by a cofilin-2 gene mutation, and this was confirmed by genetic studies. The case illustrates the ultrastructural variability that may be encountered within the nemaline myopathies and highlights the importance of electron microscopy in providing morphological correlation with genetic studies.

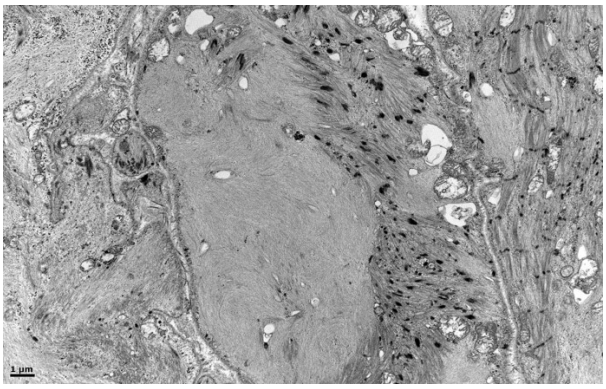

Figure 1. Abundant nemaline rods and a large filamentous aggregate within a diseased myofibre.

References

1. Sewry C, Laitila J, Wallgren-Pettersson C. Nemaline myopathies: a current view. J Muscle Res Cell Motil (2019) 40:111–126.