Nemaline Rods: A Robust Entity Surviving Paraffin Reprocessing in Fetal Autopsy Muscle

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Abstract

Background

Congenital Nemaline Myopathies display a broad range of clinical phenotypes, from severe neonatal presentations to milder forms appearing in childhood. Mutations in α-actin (ACTA1) and nebulin (NEB) are the most common causes. The rod bodies can be identified on resin sections with light microscopy, and electron microscopy confirms a distinctive lattice ultrastructure. Most muscle biopsies are on performed on live infants and children. We were unsure whether nemaline rods could be detected in fetal autopsy muscle.

Case Report

A 15-week fetus with a homozygous NEB mutation and severe arthrogryposis underwent termination. The limited autopsy allowed a muscle biopsy only from the proximal lower limb to look for nemaline rods. Unfortunately, the frozen section and glutaraldehyde-fixed sample lacked skeletal muscle. The paraffin section contained skeletal muscle and was reprocessed for electron microscopy. We identified rods on the resin sections, and typical nemaline rods ultrastructurally.

Discussion

Reprocessing paraffin-embedded tissue for electron microscopy damages membranes. However, nemaline rods are protein, and maintain their ultrastructural morphology, even in reprocessed paraffin-embedded autopsy tissue. We confirmed nemaline rods can be detected in fetal autopsy muscle.

References

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