What is in the Myopathy Literature?

Lacomis D

Departments of Neurology and Pathology (Neuropathology), University of Pittsburgh School of Medicine, Pittsburgh, PA.

Abstract

In this issue, an article describing a newly defined entity, myoglobinopathy, is covered. This autosomal-dominant, adult-onset, proximal-predominant myopathy may be associated with cardiac involvement and is due to a mutation in MB. The presence of sarcoplasmic bodies is distinctive in muscle biopsy specimens. Next, variability in phenotypes and genotypes in patients with RYR1 and TTN mutations is described. Several articles address respiratory dysfunction in myotonic dystrophy type 1, reporting that its severity is associated with the CTG-repeat size, age, and degree of muscle weakness. Several articles focus on muscle pain, including myalgias in mitochondrial disorders and the presence of inflammation in muscle biopsy specimens from patients with myalgias and abnormal electrodiagnostic testing. Finally, a form of 3-hydroxy-3-methylglutaryl-coenzyme A reductase (HMGCR) myopathy mimicking limb-girdle muscular dystrophy is highlighted.

PMID: 31453849 DOI: 10.1097/CND.000000000000261