Antibodies to Clustered AChRs and MG

Rodríguez Cruz and colleagues determine the diagnostic usefulness of cell-based assays (CBAs) in the diagnosis of myasthenia gravis (MG) and compare the clinical features of patients with antibodies only to clustered acetylcholine receptors (AChRs) with those of patients with seronegative MG. Radioimmunoprecipitation assay (RIPA) and CBA were used to test for standard AChR antibodies and antibodies to clustered AChRs in 138 patients. Cell-based assay is a useful procedure in the routine diagnosis of RIPA-negative MG, particularly in children, and patients with antibodies only to clustered AChRs appear to be younger and have milder disease than other patients with MG. Editorial perspective in support of these data is provided by Steven Vernino, MD, PhD.

Biogenesis and Myogenesis in SMA

Ripolone and coauthors investigate mitochondrial dysfunction in a large series of muscle biopsy samples from patients with spinal muscular atrophy (SMA). They studied quadriceps muscle samples from 24 patients with genetically documented SMA and paraspinal muscle samples from 3 patients with SMA-II undergoing surgery or scoliosis correction. Their results strongly support the conclusion that an altered regulation of myogenesis and a downregulated mitochondrial biogenesis contribute to pathologic change in the muscle of patients with SMA.

ACTA1 Mutation in Scapuloperoneal Myopathy

Zukosky et al determine the genetic cause of a slowly progressive, autosomal dominant, scapuloperoneal neuromuscular disorder by using linkage and exome sequencing. Fourteen affected individuals in a 6-generation family with a progressive scapuloperoneal disorder were evaluated. This family defines a new scapuloperoneal phenotype associated with an ACTA1 mutation.

Brain Stimulation for Torsion Dystonia

Fox and Alterman review the evidence and effect sizes for treating different types of dystonia with different types of brain stimulation and discuss recent advances relevant to patient selection, surgical approach, programming, and mechanism of action. They report that strong (level B) evidence supports the use of deep brain stimulation (DBS) for the treatment of primary generalized or segmental dystonia, especially when due to mutation in the DYT1 gene, as well as for patients with cervical dystonia. Patients with dystonia that is not adequately controlled with standard medical therapy should be referred for consideration of DBS, especially patients with generalized, segmental, or cervical dystonia.