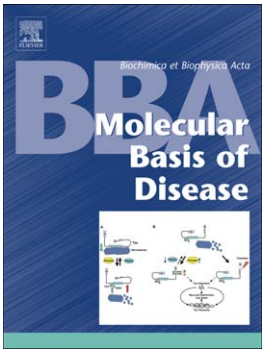

Update on Neuromuscular Diseases: Pathology and Molecular Pathogenesis

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Update on Neuromuscular Diseases: Pathology and Molecular Pathogenesis

Co-Editors: Valerie Askanas and W. King Engel

Neuromuscular Diseases encompass cellular disorders of the **Motor-Unit**, which include the *lower-motor neurons* in the spinal-cord, peripheral-nerve *schwann cells*, *neuromuscular junctions*, and *skeletal muscle fibers*.
Neuromuscular disorders can be primary, or consequent to another illness, such as cancer, collagen-vascular disorder, or a generalized autoimmune, metabolic, deficiency infectious or toxicity disorder, and they are disabling millions of persons. Moreover, muscle weakness and frailty with ageing are virtually inevitable, generally beginning to become manifest beyond the age of 40 – they progress to various degrees, and are unstoppable. They can lead to reduced mobility, increased risk of falling, injury, paralysis, and even death.
In Neuromuscular Diseases, the *motor component* i*nvolvement* causes muscle weakness and atrophy, resulting in falling and various degrees of paralysis, including involvement of limbs, breathing, swallowing, and, infrequently, eye movements. All are troublesome, frustrating and crippling, and some are dangerous and/or rapidly fatal. Motor involvement also can cause painful muscle cramps and spasms. Involvement of the *sensory component* of the neuromuscular system causes pain, numbness, tingling, incoordination of limbs, imbalance and falling.

For neuromuscular diseases there is an abundant efflorescence of new information very-cogent to: a) diagnosis and understanding of pathogenesis (e.g. genetic mutations, characteristic muscle biopsy abnormalities, and various specific biomarkers); and b) newly-discovered steps of pathogenic pathways leading to formulation of current and future treatments (e.g., disease-specific targeted therapy). Recent clarification of genetic abnormalities involved in several hereditary neuromuscular diseases has greatly added to our understanding of molecular mechanisms involved in them, as well as in several relevant sporadic neuromuscular diseases.

The newest, fascinating details of examples in each category are presented by experts.