INHERITED NEUROMUSCULAR DISEASES

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The main feature of neuromuscular disorders is muscle weakness. Several other associated muscle characteristics, like cramps, muscle slowness and relaxation disturbances, enable to narrow down the vast amount of neuromuscular diagnostic possibilities. In addition, these features give insight into pathophysiological and physiological mechanisms.

These concepts will be illustrated by three genetically defined neuromuscular diseases: 1) Non-dystrophic myotonia, 2) Brody's disease and 3) Nemaline myopathy. Non-dystrophic myotonia's are genetically defined chloride- or sodiumchannelopathies of the muscle membrane giving rise relaxation disturbances after rest and a sometimes transient paresis. Brody's disease is a disease of the sarcolemmal CaATPase causing a relaxation disturbance during exercise. Nemaline myopathies typically seem to affect the thin filaments as the 5 genes so far identified all encode for components of the thin filaments. We demonstrate slow muscle characteristics as a physiological correlate for the clinical complaint of slowness in movement.

In conclusion, these three groups of genetically defined muscle disorders (involving a muscle membrane channel, CaATPase activity, thin filament) illustrate various mechanisms of muscle slowness in relaxation and in contraction.

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