# Pediatric Neurology Part III: Chapter 147. Muscle channelopathies and related diseases (Handbook of Clinical Neurology)

*Bertrand Fontaine*

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Muscle channelopathies and related disorders are neuromuscular disorders predominantly of genetic origin which are caused by mutations in ion channels or genes that play a role in muscle excitability. They include different forms of periodic paralysis which are characterized by acute and reversible attacks of muscle weakness concomitant to changes in blood potassium levels. These disorders may also present as distinguishable myotonic syndromes (slowed muscle relaxation) which have in common lack of involvement of dystrophic changes of the muscle, in contrast to dystrophia myotonica. Recent advances have been made in the diagnosis of these different disorders, which require, in addition to a careful clinical evaluation, detailed EMG and molecular study. Although these diseases are rare, they deserve attention since patients may benefit from drugs which can dramatically improve their condition. Patients may have atypical presentations, sometimes life-threatening, which may delay a proper diagnosis, mostly in the first months of life. The creation of specialized reference centers in the Western world has greatly benefited the proper recognition of these neuromuscular diseases.

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