Pediatric Neurology Part III: Chapter 144. Congenital and infantile myotonic dystrophy (Handbook of Clinical Neurology)

Bernard Echenne, Guillaume Bassez

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Myotonic dystrophy (DM) encompasses two gene defects, DM1 (myotonic dystrophy type 1) being currently the sole disorder leading to a childhood form of the disease. As consequence of the non coding unstable CTG repeat expansion mutation, DM1 presents as an extremely wide and diverse clinical continuum ranging from antenatal to late adult forms, the complexity of the disease being reinforced by multisystemic involvement. The congenital form appears as the most severe end of the phenotypic spectrum and may include marked neonatal hypotonia, respiratory failure, facial diplegia, contractures, and mental retardation. Brain involvement is the hallmark of childhood-onset DM1, distinguished by a normal neonatal period, with learning difficulties as the main presenting symptom, resulting from various degrees of mental delay, psychopathological manifestations, speech defects, hypersomnolence, and fatigue. In contrast, muscle weakness remains usually moderate in childhood, limited to facial weakness, ptosis, and dysarthria, until a decline from the second decade. Orthopedic manifestations including kyphoscoliosis and equinovarus may require surgery. Other organs involvement includes frequent abdominal symptoms, whereas endocrine disturbance is rare. Symptomatic cardiac arrhythmia, mainly exercise-induced, can be observed. While current treatment is mainly symptomatic, future clinical trials are expected following significant progress in pathophysiology and the recent development of molecular therapy approaches.

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