



POSNA

The Core Curriculum

Congenital myopathies

Objectives

1. Describe clinical features which accompany congenital myopathies
2. Discuss pertinent anesthetic risk factors when considering surgery for congenital myopathies

Discussion

The congenital myopathies comprise a group of disorders characterized by hypotonia and weakness. Central core disease is a nonprogressive autosomal dominant with muscle weakness similar to Duchenne muscular dystrophy, except that weakness accompanying central core disease is easily detectable in infancy and motor development is delayed. Diagnosis is established by muscle biopsy. Scoliosis, soft tissue contractures, clubfeet, and hip dislocation have been associated with central core disease. Nemaline rod myopathy (moderate congenital form) is characterized by delayed motor milestones, and occasional spinal deformity. Bracing may be helpful to achieve ambulation. Myotubular myopathy, sometimes called centronuclear myopathy, has variable genetic patterns of transmission and variable phenotypes. High arched palates occur with both nemaline rod and myotubular myopathies. Some children with myotubular disorders may appear phenotypically similar to myotonic dystrophy. Congenital fiber type disproportion is manifest as hypotonia at birth by hypotonia. Muscle biopsy demonstrates a deficiency of type I fibers. The degree of weakness is variable.

Central core disease and nemaline rod neuropathy are associated with an increased risk of malignant hyperthermia during anesthesia.

One can expect clarification in this group of disorders with molecular biology studies, but to date this group of disorders is characterized by considerable phenotypic variation.

References

1. Baccetti T, Defraia E, Donati MA. Craniofacial abnormalities associated with congenital fiber type disproportion myopathy. *Journal of Clinical Pediatric Dentistry* 1997;21(2):167-71.
2. Danon MJ, Giometti CS, Manaligod JR, Swisher C. Sequential muscle biopsy changes in a case of congenital myopathy. *Muscle & Nerve* 1997;20(5):561-9.

3. De Angelis MS, Palmucci L, Leone M, Doriguzzi C. Centronuclear myopathy: clinical, morphological and genetic characters. A review of 288 cases. *Journal of the Neurological Sciences* 1991;103(1):2-9.
4. Estournet-Mathiaud B. Respiratory problems in congenital myopathies. *Pediatric Pulmonology - Supplement* 1997;16:142.
5. Kim WK, Choi BO, Cheon HY, Sunwoo IN, Kim TS. Muscle fiber type disproportion with an autosomal dominant inheritance. *Yonsei Medical Journal* 2000;41(2):281-4.
6. Nonaka I, Ishiura S, Arahata K, Ishibashi-Ueda H, Maruyama T, Ii K. Progression in nemaline myopathy. *Acta Neuropathologica* 1989;78(5):484-91.
7. Patterson MC, Gomez MR. Muscle disease in children: a practical approach. *Pediatrics in Review* 1990;12(3):73-82.
8. Rowe PW, Eagle M, Pollitt C, Bullock RE, Bushby KM. Multicore myopathy: respiratory failure and paraspinal muscle contractures are important complications. *Developmental Medicine & Child Neurology* 2000;42(5):340-3.
9. Shapiro F, Specht L. The diagnosis and orthopaedic treatment of inherited muscular diseases of childhood [see comments]. *Journal of Bone & Joint Surgery - American Volume* 1993;75(3):439-54.
10. Thompson GH. Neuromuscular disorders. In: Morrissy RT, Weinstein SL, editors. *Pediatric Orthopaedics*. Philadelphia: Lippincott-Raven Press; 1996. p. 537-77.