
A survey of 556 pediatricians in Virginia revealed an overall deficiency in physicians' knowledge of lead poisoning with specific deficiencies in knowledge of the literature. Subspecialists scored lower than primary care pediatricians. (Bar-on ME, Boyle RM. Are pediatricians ready for the new guidelines on lead poisoning? Pediatrics Feb 1994;93:178-182). Since 17% of all children <7 years in the USA are reported to have elevated BPb levels known to increase the risk of cognitive and behavioral deficits, both physician and parent awareness of the environmental sources, symptoms, and long-term hazards of lead is an urgent priority. (Millichap JG. Environmental Poisons in Our Food, Chicago, PNB Publishers, 1993).

MUSCLE DISORDERS

CONGENITAL NEMALINE MYOPATHY

A female neonate with a rapidly fatal course of nemaline myopathy is reported from the University of Siena, Italy. Positive pressure ventilation was required and postasphyxia suspected. Despite improved cardiorespiratory function, severe hypotonia, muscle weakness and areflexia persisted. At 2 months, fractures of both femurs and left humerus were noted, and a myopathy was considered in diagnosis. Muscle biopsy of quadriceps showed rod-shaped nemaline bodies. The infant died at 4 months of pneumonia. Nemaline bodies were found in diaphragm, intercostal, psoas, and quadriceps muscles. The heart was also involved. The parents were healthy and their muscle biopsies normal. (Buonocore G et al. A new case of severe congenital nemaline myopathy. Acta Paediatr Dec 1993;82:1082-4). (Respond: Dr G Buonocore, Division of Neonatology, University of Siena, via P Mascagni, 46 53100 Siena, Italy).

COMMENT. Persistence of severe hypotonia in a neonate, together with dependence on assisted ventilation, should prompt investigation of a possible myopathy.

Intranuclear rods were present in muscle fibers of one infant with a rapid, fatal course of nemaline myopathy but were absent in the muscles of seven patients with a benign course, in a study reported from the Departments of Neurology and Pathology, University of Rochester Medical Center, NY. (Rifai Z et al. Intranuclear rods in severe congenital nemaline myopathy. Neurology Nov 1993;43:2372-2377). The presence of intranuclear rods represents a marker for the severe form of congenital nemaline myopathy.

The clinical manifestations of three forms of nemaline myopathy are reported as follows: 1) severe neonatal form, with hypotonia, feeding and respiratory difficulties, and death in infancy; 2) nonprogressive or slowly progressive form, presenting in infancy or early childhood with delayed motor milestones and facioscapuloperoneal weakness; and 3) adult-onset form, with a progressive proximal weakness. The term "congenital nemaline myopathy" is applied to forms 1) and 2). The authors caution that the neonatal type is not invariably fatal, and improvement may occur, or alternatively, deterioration may follow an initial stable course.