PEDIATRIC NEUROLOGY BRIEFS

A MONTHLY JOURNAL REVIEW

J. GORDON MILLICHAP, M.D., F.R.C.P., EDITOR

Vol. 2, No. 9 September 1988

NEONATAL NEUROMUSCULAR DISORDERS

NEONATAL RHABDOMOLYSIS AND DYSTROPHY

A boy, aged 2 years, presenting at birth as a case of rhabdomyolysis and later shown to have an X-linked recessive dystrophy, is reported from the Section of Child Neurology, St. Christopher's Hospital for Children, Philadelphia, PA, and the Child Neurology A.I. Dupont Institute, Wilmington, DE. Palpation of upper and lower extremities on newborn examination revealed stiff, indurated large muscle groups. The birth was a cephalic presentation without forceps. The CK at 2 days of age was 156,000 IU/l, and a benzidene dipstick for heme and myoglobinuria was negative at 4 days. Repeat CK determinations at 6 days and between 5 weeks and 14 months were approx. 12,000 and 6000-9000 IU/l, respectively. Percutaneous needle biopsy of the quadriceps at 1 year demonstrated many degenerating fibers, marked variation of fiber size, and increase in endomysial and perimysial connective tissue and fat. DNA analysis showed a partial X chromosome deletion adjacent to the Duchenne/Becker locus. On clinical examination the infant was developmentally delayed at 11 months and had speech delay, proximal lower extremity weakness, and calf pseudohypertrophy at 24 months. (Breningstall GN et al. Neonatal rhabdomyolysis as a presentation of muscular dystrophy. Neurology Aug 1988;38:1271-1272).

COMMENT. Rhabdomyolysis is an acute muscle necrosis usually accompanied by myoglobinuria which may complicate

PEDIATRIC NEUROLOGY BRIEFS © 1988 covers selected articles from the world literature and is published monthly. Subscription requests ($28 US or £ 15 UK annually; add $5 (£3) for airmail outside North America) may be sent to: Pediatric Neurology Briefs - J. Gordon Millichap, MD, FRCP - Editor, P.O. Box 11391, Chicago, IL 60611, USA, or P.O. Box 1605, London W8 7JD, UK.

The Editor, Professor of Neurology and Pediatrics at Northwestern University Medical School, Chicago, is presently at SIU School of Medicine, Springfield, Illinois, USA.

muscular dystrophy in association with malignant hyperthermia or anesthetic induced cardiac arrest. This patient had neither myoglobinuria nor hyperthermia. A neonatal presentation of Duchenne muscular dystrophy is exceptional, signs usually appearing when the patient becomes ambulant.

NEONATAL GUILLAIN-BARRE' SYNDROME

A term female infant presenting with generalized hypotonia, paucity of lower limb movements, and diminished DTRs was diagnosed as a case of Guillain-Barre' syndrome at the Hospital for Sick Children, University of Toronto, Canada. At 3 weeks of age, motor nerve conduction studies showed slowed velocities and decreased action potentials, and the CSF protein was elevated with normal cells. Clinical improvement began at age 2 weeks and examination was normal at 22 weeks. (Al-Qudah AA et al. Neonatal Guillain-Barre' syndrome. Pediatr Neurol Aug 1988;4:255-6).

NEONATAL MYASTHENIA GRAVIS

The advantages of electrodiagnosis in a premature infant with neonatal myasthenia gravis are proposed by the Dept of Rehabilitation Medicine, Children's Hospital and Medical Center, Univ of Washington School of Medicine, Seattle, WA. The infant, born to a mother with myasthenia, suffered hypoxia and subependymal hemorrhage which probably contributed to the hypotonia and poor respiratory effort. Testing with edrophonium, 0.1 mg/kg IV demonstrated no clinical improvement, whereas repetitive motor nerve stimulation testing showed a significant decremental response consistent with a diagnosis of neonatal myasthenia gravis. The decremental response was corrected following IV infusion of edrophonium 0.15 mg/kg. Pyridostigmine in a dose of 8 mg/kg/day resulted in clinical improvement of respiratory and muscular activity, and the infant was weaned from assisted ventilation at 27 days of age. He was discharged at 41 days of age on pyridostigmine therapy. The authors conclude that repetitive motor nerve stimulation may be a more reliable diagnostic procedure than edrophonium IV in the newborn with suspected myasthenia gravis. (Hays RM, Michaud LJ. Neonatal myasthenia gravis: Specific advantages of repetitive stimulation over edrophonium testing. Pediatr Neurol Aug 1988;4:245-7).

COMMENT. The value of electrodiagnostic tests in the differential diagnosis of the hypotonic infant is demonstrated in these 2 case reports. Neonatology texts often recommend edrophonium as the test of choice in neonatal myasthenia gravis. The above experience indicates that the pharmacological test alone may not be as sensitive as repetitive nerve stimulation in the newborn with multiple problems. Ultrasonography is an additional technique of potential value in the work up of the hypotonic infant. Heckmatt JZ and Dubowitz V of