NEUROMUSCULAR DISORDERS

FAMILIAL MYOPATHY WITH THROMBOCYTOPENIA

Three cases of a familial myopathy with thrombocytopenia in 3 generations of a family are reported from the Royal Manchester Children's Hospital, Withington Hospital, and University of Manchester Medical School, England. The family showed autosomal dominant inheritance of both myopathy and thrombocytopenia. The myopathy presented in early childhood with an abnormal gait noted by the age of 4 years and an asymmetric limb girdle weakness diagnosed in the proband by 8 years. By puberty, distal upper limb weakness had developed, and the myopathy was slowly progressive as evidenced by increasing disability in the mother and wheelchair confinement in the grandfather. The asymmetrical myopathic findings became generalized with increasing age. The CK was elevated in all 3 patients (800, 600, and 524 IU/l). EMG obtained in 2 patients was normal in the son at 9 years, and showed a high amplitude motor unit with a slightly diminished interference pattern in the mother at 32 years of age. The bleeding disorder manifested by easy bruising and prolonged bleeding following injury was fully expressed in the proband when first examined at 8 years and the platelet disorder showed no progression with age. Muscle biopsies from mother and son showed type I fiber preponderance and hypertrophy, and type II fiber atrophy and rimmed vacuoles. Electron-microscopy revealed tubular aggregates in both cases. (Mahon M et al. Familial myopathy associated with thrombocytopenia: a clinical and histomorphometric study. J Neurol Sci Dec 1988;88:55-67).

> **COMMENT.** Limb girdle dystrophy is not a single entity and should prompt examination of the blood for platelet abnormalities. This appears to be the first recorded family with defects of both muscle and platelets in the same patients.

FAMILIAL MYOPATHY WITH INCLUSION BODY MYOSITIS

Five male siblings affected by a progressive myopathy, inclusion body myositis, and periventricular leukoencephalopathy are reported as a new syndrome from the Montreal Neurological Hospital and Institute, Canada. Patient 1, the first of twins, examined at 35 years of age, walked at 18 months but was never able to run. Muscle weakness, mainly proximal, progressed slowly during adolescence, and a cane was required to walk by 26 years. At 35 years, he had genu recurvatum and excess lumbar lordosis, proximal weakness with minimal wasting, and absent tendon jerks in the upper limbs, reduced at the knees, and preserved at the ankles. He required crutches to walk, and he had a waddling gait. His mentation, cranial nerves, sensation, and coordination were normal. The 4 siblings had similar histories and findings. CK was elevated, EMG showed small polyphasic motor units, fibrillation, and positive sharp waves; NCV showed minimal slowing; CSF protein was 0.56-0.68 g/l; and muscle biopsies revealed fiber loss, rimmed vacuoles, variability of fiber calibre, and some necrosis, and abnormal

cytoplasmic filamentous inclusions. CT white matter hypodensities and MRI high signal intensities were compatible with leukodystrophy, yet the patients had no symptoms of white matter dysfunction. The authors conclude that this constellation of familial myopathy with muscle cytoplasmic inclusions and cerebral white matter changes represents a hitherto undescribed syndrome. (Cole AJ et al. Familial myopathy with changes resembling inclusion body myositis and periventricular leucoencephalopathy. Brain Oct 1988;111:1025-1037).

> **COMMENT.** Familial cases of inclusion body myositis have been reported in younger patients but none associated with cerebral white matter changes as described above. In contrast to those myopathies sometimes associated with CNS dysfunction (eg. Duchenne muscular dystrophy, myotonic dystrophy, congenital muscular dystrophy of Fukuyama and others, and Kearns-Sayre-Shy syndrome) the white matter changes in the present cases were asymptomatic. The mode of inheritance could not be determined with certainty. Limb girdle dystrophy presents in yet another form and should prompt examination not only of blood platelets but also CT and MRI for white matter changes. CT findings were not included in the report from Manchester and blood platelet counts were not indicated in the Montreal syndrome.

CONGENITAL MYOPATHY, CLEFT PALATE, AND MALIGNANT HYPERTHERMIA

Six children with congenital ptosis, generalized weakness, hypotonia, cleft palate, and susceptibility to malignant hyperthermia with anesthesia, are reported in Lumbee Indians from Duke University Medical Center, Durham, North Carolina. All patients were members of the same ethnic group, 3 were related, and inheritance was probably autosomal recessive. Surgery for cleft palate at 14 months and for ptosis at 27 months, using halothane anesthesia, was complicated by malignant hyperthermia in one child. This syndrome showed some resemblance to King syndrome, characterized by multiple congenital facial and skeletal abnormalities along with slowly progressive myopathy and susceptibility to malignant hyperthermia. (Stewart CR et al. Congenital myopathy with cleft palate and increased susceptibility to malignant hyperthermia: King syndrome? Pediatr Neurol Nov/Dec 1988;4:371-4).

> **COMMENT.** Clinicians, especially surgeons and anesthesiologists, should be aware of the risk of malignant hyperthermia in children with this syndrome and other myopathies, including Duchenne muscular dystrophy, myotonia congenita, and central core disease. Malignant hyperthermia is manifested by muscle rigidity, rapid elevation of temperature, metabolic acidosis, and rhabdomyolysis. Anesthetic agents most frequently invoked are halothane and succinylcholine. Screening of susceptible patients and their families by CK determinations is advised.

An index for PEDIATRIC NEUROLOGY BRIEFS, Vols 1 and 2, 1987-88 is in preparation.