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