NEUROMUSCULAR DISORDERS

PERIPHERAL NEUROLOPATHY IN XERODERMA PIGMENTOSUM

The peripheral nerve pathology in two autopsied cases of group A xeroderma pigmentosum (De Sanctis Cacchione syndrome) is reported from the Tokyo Medical and Dental University, Tokyo Metropolitan Neurological Hospital, and Tokyo Metropolitan Kita Medical and Rehabilitation Center, Tokyo, Japan. One patient died at 19 years of age because of intractable respiratory tract infection and acute renal failure and the other died aged 23 due to choking. The diagnosis of xeroderma pigmentosum had been made in early infancy because of prominent light sensitivity. Neurological symptoms had developed in childhood and included slurring of speech, ataxia, and mental retardation. The patients were bedridden at age 15 and 22. Examination revealed microcephaly, short stature, and hyperpigmentation of the skin exposed to sunlight. There was severe muscle atrophy in all limbs, contractures of joints, and fasciculation of the tongue. Tendon reflexes were absent and plantar responses were extensor. Evoked muscle action potentials and sensory action potentials were absent on stimulation of peripheral nerve trunks. Pathologic changes in the nerves suggested a neuronopathy with loss of myelinated nerve fibers and endoneurial fibrosis. Changes in the spinal cord included a severe decrease in anterior horn cells, reduction in lateral columns and severe depletion of dorsal root ganglion cells. The brains were small and showed widespread sclerotic leucoencephalopathy and severe neuronal loss in the cerebral cortex, thalamus, substantum nigra and cerebellar cortex. (Kanda T et al. Peripheral neuropathy in xeroderma pigmentosum. Brain August 1990; 113:1025-1044).

> COMMENT. Xeroderma pigmentosum is a group of autosomal recessive disorders related to a defect in the mechanism of DNA repair. The findings in the peripheral nerves are similar to those reported in ataxia telangiectasia, another disorder which shows defective DNA repair. The authors suggest a common pathogenic mechanism.

PERIPHERAL NEUROPATHY IN ATAXIA-TELANGIECTASIA

EMG examinations performed on 32 children aged three to 13 years with ataxia-telangiectasia are reported from the Neurological Department of the Child Health Centre, Warsaw, Poland. Four main EMG patterns were distinguished: 1) normal, 2) increased polyphasia of motor unit potentials, 3) neurogenic lesions with denervation activity, 4) denervation, fasciculations, and a picture characteristic of advanced motor neuron disease. The severity of the neurogenic lesions increased from the proximal arm muscles to the distal leg muscles. An EMG pattern resembling motor neuron disease was seen most often in the extensor digitorum brevis. Nerve conduction studies showed a decrease in motor response amplitude in the older children and a reduction in sensory nerve action potentials in median and sural nerves of all children older than seven years. The authors consider that a generalized slowly progressive sensory system degeneration together with neurogenic amyotrophy affecting distal parts of the

lower limbs is a constant feature of ataxia telangiectasia and can be taken to be one of the diagnostic characteristics. (Kwast O, Ignotowicz R. Progressive peripheral neuron degeneration in ataxia-telangiectasia: An electrophysiological study in children. Dev Med Child Neurol September 1990; 32:800-807).

COMMENT. The clinical diagnosis of ataxia-telangiectasia is based on a progressive cerebellar ataxia, ocular telangiectasia, and immunological abnormalities. Muscle weakness progresses with age and atrophy affects especially the distal leg muscles. The child is confined to a wheelchair after the 9th to the 12th year. EMG and nerve conduction studies are important in the diagnosis.

DYSTROPHIN AND DIAGNOSIS OF MUSCULAR DYSTROPHY

The value of dystrophin analysis in the early diagnosis of two patients with childhood autosomal recessive muscular dystrophy is reported from the Department of Pediatrics, Sapporo Medical College, Sapporo, Japan. The first patient, a five year old boy referred because of an elevated serum CK had developed normally until two years of age when an abnormal gait was observed. He had slight proximal muscle weakness without enlargement of calf muscles or involvement of facial muscles. Gower's sign was negative. The deep tendon reflexes in the lower limbs were slightly decreased. A muscle biopsy from the biceps showed marked variation in fiber size and a number of necrotic and regenerating fibers with proliferating connective tissue characteristic of muscular dystrophy. Dystrophin was demonstrated in all sarcolemma after immunocytochemical staining using antidystrophin antibody. The second patient, a seven year old boy with rubella, was found to have an elevated serum CK. His motor milestones of development were normal and neither muscular weakness nor atrophy were observed. There was no enlargement of calf muscles or involvement of facial muscles. Deep tendon reflexes were slightly hypoactive. The serum CK was 4250 IU. From seven to 11 years of age the clinical course was static. Muscle biopsies from the rectus femoris and biceps brachii showed a marked variation in fiber size and degenerating and regenerating fibers with proliferated connective tissue indicative of muscular dystrophy. By immunocytochemical staining with antidystrophin antibody the dystrophin was located in the sarcolemma. Both patients were diagnosed as having childhood autosomal recessive muscular dystrophy. There was neither consanguinity nor any history of neuromuscular disorder in the families. (Tachi N et al. Dystrophin analysis in the differential diagnosis of autosomal recessive muscular dystrophy of childhood and Duchenne muscular dystrophy. Pediatr Neurol July-August 1990; 6:265-268).

COMMENT. In boys with a muscular dystrophy that develops in early childhood the early differentiation of Duchenne, Becker, and the autosomal recessive limb-girdle dystrophies is important. The onset of limb-girdle dystrophy is more commonly between 10 and 20 years of age but it may present in the first decade and sometimes as early as two years of age. The course is variable but occasionally progression is as rapid as with Duchenne muscular dystrophy.