

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, substantia 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