HEREDO-DEGENERATIVE DISEASES

X-LINKED ATAXIC SYNDROME

An X-linked recessive disease with a fatal course in early childhood is reported in a five-generation Dutch family from the Netherlands. Twelve boys were affected and 13 female carriers were identified, some with hearing impairment. Neurological deterioration coincided with recurrent respiratory infections at 1 year of age or earlier and was characterized by hypotonia, ataxia, weakness, absent deep tendon reflexes, nystagmus, and visual and hearing loss. Autopsy in one patient revealed absence of myelin in posterior columns of the spinal cord, and axonal degeneration in the peripheral nerves. The brain was normal in appearance. No biochemical or immunological defects were detected. (Arts WFM et al. X-linked ataxia, weakness, deafness, and loss of vision in early childhood with a fatal course. Ann Neurol May 1993; 33: 535-539). (Respond: Dr Arts, Dept of Neurology, Westeinde Hospital, PO Box 432, 2501 CK The Hague, The Netherlands).

> COMMENT. Mitochondrial encephalomyelopathy and other known metabolic or degenerative diseases were excluded. The autopsy findings in one patient resembled those of Friedreich's ataxia, but the age of onset and early death were atypical.

ALPERS SYNDROME WITH HEPATIC CIRRHOSIS

Four children, from two families, with fatal degeneration of the cerebral grey matter and terminal hepatic dysfunction are reported from the Royal Belfast Hospital for Sick Children, Northern Ireland. The disease presented with intractable generalized or partial seizures during infancy in 3 and at 5 years of age in one patient. Epilepsia partialis continua was associated with ataxia and progressive neurologic deterioration. The EEG showed high amplitude slow waves with smaller poyspikes. Visual evoked responses were delayed, and CTs showed cerebral atrophy. Post mortem findings in one patient included neuronal loss and gliosis and hepatic cirrhosis. (Wilson DC et al. Progressive neuronal degeneration of childhood (Alpers syndrome) with hepatic cirrhosis. Eur J Pediatr 1993; 152(3): 260-262). (Respond: Dr DC Wilson, Neonatal Unit, Royal Maternity Hospital, Grosvenor Road, Belfast BT126BJ, N Ireland).

> COMMENT. Valproic acid, sometimes implicated in deaths of young infants with this syndrome, was apparently not a factor in these cases. Early diagnosis allows appropriate genetic counselling.

MITOCHONDRIAL MYOPATHY AND CONGENITAL CATARACT

The autosomal recessive syndrome characterized by mitochondrial myopathy of cardiac and skeletal muscle, congenital cataract and lactic