Infantile Neuroaxonal Dystrophy:
Cortical Axonic and Presynaptic Changes

U. SANDBANK, P. LERMAN and M. GEIFMAN
J. Casper Institute of Pathology, Laboratory of Neuropathology, E.E.G. laboratory and the Department of Pediatrics Beilinson Hospital and the Tel Aviv University Medical School

Received June 1, 1970

Summary. A 2½ year old girl who, since the age of 1 year presented, a progressive psychomotor retardation. A cortical biopsy appeared normal by light microscopy, but by electron microscopy an abundance of dilated spheroid-like axons were found. They contained either vesiculo-tubular material or densely packed filamentous material. Synapses were demonstrated between the spheroids and other neuronal bodies or dendrites. Crystalline like material was observed within mitochondria and the spheroids. It is suggested that Neuroaxonal dystrophy may be diagnosed by cortical brain biopsy.

Key-Words: Neuroaxonal Dystrophy, Infantile — Cortical Biopsy — Spheroid-Like Axons — Electron Microscopy.

The presence of large spheroidal axonic swellings is the typical morphological lesion found in Infantile Neuroaxonal Dystrophy (I.N.A.D.). Recently, reports describing the electron microscopic findings of I.N.A.D. cases (Hedley-Whyte et al., 1968; Herman et al., 1969); stress the possibility of diagnosis by cortical biopsy. The present case describes a child with progressive psychomotor degenerative disease of the nervous system with electron microscopical findings in a cortical biopsy of axonic and presynaptic changes suggestive of I.N.A.D.

Case Report

A 2½ year old Arab girl was referred with progressive weakness, starting at the age of 11—12 months. The parents, who were second cousins, were in good health. They had previously had 2 normal children. A younger child, however, was found soon after birth to be retarded and eventually died at the age of 1 year. An autopsy was not performed.

The patient was the product of a normal, full term pregnancy. Birth weight was 3,300 g. Her initial development was normal: she sat up by 5—6 months, stood by 8 months, and started talking and walking with support by 11 months. However, from the age of 1 year there was evidence of regression—she stopped talking and gradually lost the ability to walk and to sit up. She was admitted to another hospital at the age of 2 years. She was found to have
muscle weakness and active deep tendon reflexes. She could not maintain a sitting position. No other physical abnormalities were found. X-rays of chest, skull and spine were normal. E.E.G and air study revealed no abnormality. The blood chemistry was normal, as were the CSF studies, including virus cultures. A muscle biopsy showed no abnormality.

At the age of 30 months the child was admitted to our hospital for further investigation. At that time she was spastic and retarded. There was head lag, she showed no evidence of understanding spoken language, and she could hardly grasp anything. She responded to sound and light. Head circumference was 46.2 cm. X-rays of the skull were normal. An EEG during

Fig. 1. Two spheroids, one containing tubulo-vesicular formations, the second consisting of fibrillar material. 17,500 x