An Unusual Type of Infantile Lipofuscinosis

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Summary. The case of a child is described who at the age of 2 years showed the first evidence of a developing neurological disease. Within a couple of years, profound mental retardation and severe motor deficit with spastic tetraplegia became established. No seizures and no pigmentation of the retina were observed. The condition remained practically unchanged for some 8 years and the patient died at 12 years of age of terminal bronchopneumonia. At autopsy there was conspicuous diffuse atrophy of the brain. The cerebral cortex was particularly involved. Most of the cortical neurons were destroyed and neuroglia showed abundant proliferation. The few remaining neurons contained inclusion material which was identified as lipofuscin. Noticeable differences from the various types of amaurotic idiocies are noted and similarities to a case of lipidosis recently reported from Finland are suggested.

Key words: Childhood -- Brain Atrophy -- Lipofuscin.

Progressive degeneration of the brain gray matter in childhood associated with clinical manifestations of mental deterioration and spastic tetraplegia has been the object of extensive studies. Aside from cases of infections, intoxication or anoxia, two distinct groups are recognized: the well defined cerebral lipoidoses (Zeman et al., 1970) and the poorly understood types described under the eponym of Alper's disease (Verhaart, 1972). The purpose of this report is to place on record an unusual type of diffuse cortical degeneration of the brain with onset at 18 months of age and progressive downhill course lasting 10 years.

Clinical Findings

M. L. a boy 6 years old, was admitted to a State institution because of profound mental retardation and quadriplegia. The unrelated parents, age 26 and 24 years, respectively, at the birth of the patient, were Germans of non-Jewish ancestry. Family history was not contributory. There were three normal sibs.

Pregnancy and delivery were normal. Development during the first year of life was uneventful. At about 18 months, hyperactivity and some difficulty in walking were noted. Shortly after, arrest of speech development, incoordination of movements and dysarthria were evident. At 4 years, mental retardation was severe, paraparesis with bilateral Babinski, hyperreflexia, unsteady gait and disorganized hyperactive behavior were reported. When admitted to our institution at the age of 5 years, the I.Q. was 15, there was spastic tetraplegia with pyramidal signs, absent abdominal reflexes, lateral nystagmus, strabismus, questionable hearing and no speech. The fundi were normal. Spleen or liver were not enlarged. Routine examination of urine, blood and spinal fluid revealed no abnormalities.

The child was followed until death. There was increase of spasticity during the first 2 years culminating into an extreme flexion contracture of all limbs and moderate extension of spine. No convulsive and no myoclonic manifestations were ever observed. Bilateral pallor of optic nerve was noted at 8 years of age, but repeated funduscopy examinations failed to detect any pigmentation of the retina. Electric retinogram was not done. During the last 4 years, the
neurological status did not change, but the general condition worsened considerably. He lost considerable weight, developed multiple decubiti and suffered several bouts of pneumonia. He died at 12 years of age of bronchopneumonia.

**Pathological Findings**

Autopsy was performed within 24 hrs of death. Apart from an acute pneumonic process and moderate fatty degeneration of the liver, no gross changes were noted in the thoracic and abdominal viscera. Permission to examine the eyes was not obtained.

The brain weighed 650 g, pia was thickened, convolutions appeared markedly atrophic and often ribbon-like (Fig. 1). All lobes were markedly involved but in the occipital lobes the medial convolutions were better preserved. Cortex was thin and often showed longitudinal splits. Lateral ventricles were markedly dilated. The white matter of the centrum ovale was considerably reduced in size and hard in consistency. Putamen and pallidus were reduced to a small formation in the lower lateral portion of the ventricle. Cerebellum and midbrain were only mildly atrophic.

![Fig.1 a and b. Severe atrophy of the brain. (a) Lateral view; (b) medial view](image-url)