Greenfield's Metachromatic Leucodystrophy
with Unusual Amount of Pigment

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With 2 Figures in the Text

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The case described below represents the late infantile form of metachromatic leucodystrophy. In spite of a certain incompleteness of material we consider its presentation relevant, owing to its distinctive properties consisting in an intracellular accumulation of pigment which seems to be independent of other products of a disturbed myelin metabolism.

Case history

A female child, Jolanta G. was admitted to the City Hospital in Opole in 1961. Up to the first year the child's development had been normal. At this time the mother noticed certain kinds of movements, the description suggesting a type of choreo-athetosis. At the age of two the child contracted severe chicken pox and her general condition underwent a steady deterioration. Six months later she was brought to the neurological department in a semicomatose state with spastic tetraparesis.

The girl died after ten days observation with diagnosis of an encephalitis.

The laboratory findings and the general autopsy data were irrelevant.

Brain microscopical findings (PAN 64/61)

In the section stained for myelin a pronounced diffuse loss of the latter was most marked in the parieto-occipital region and in the centrum semiovale. There was a complete demyelination of the external capsules and of the basal ganglia fibres system. In Holzer's preparations the discoloured, demyelinated parts corresponded to a particulary dense fibrillary gliosis. On the frozen sections, in ordinary cell staining, it looked as if the whole of the white matter was totally devoid of glial nuclei, but indeed only the oligodendroglia had disappeared. In the sections stained with acetic cresyl-violet the white matter was packed with brown metachromatic deposits, stored in the swollen, often binucleated astrocytes and relatively few macrophages. The storage of the metachromatic material could be seen in the cortical neurons as well as in the subcortical ganglia.

In the claustrum and in the outer part of the putamen the extensive perivascular lacunae with sponzous rarefaction of the nervous tissue were seen.

A most characteristic feature of this case was the presence of a peculiar pigment accumulated in the cytoplasm of astrocytes, neurons, perivascular macrophages and vascular endothelium. The amount of this pigment was especially abundant in the external capsule, claustrum, outer part of the putamen and in the vicinity of the subependymal area around the occipital horn of the lateral ventricle. In those places where its accumulation was greatest it was apparent to the naked eye in unstained sections. In hematoxyline-eosine stained preparations the pigment was partly yellow and amorphous, partly in the form of dark-brown and black-gold crystals (Figs.1 and 2). It was black in Sudan black B, staining both in paraffin and frozen section and red in PAS staining, but negative in Hirsch-Peiffer test. Negative results were obtained also with Sudan III, PFAS, copper phthalocyanin, Alcian blue, toluidine blue, Feyrter and Turnbull blue stainings.
Metachromatic leucodystrophy with unusual amount of pigment

Discussion

The general morphological and chemical features of this case correspond to Greenfield's type of leucodystrophy, which nowadays is distinguished by a well defined position (JERVIS 1960; SOURANDER AND SVENNERHOLM 1961; NORMAN 1963).

Fig. 1. Occipital lobe: pigment in astrocytes in subependymal area. Paraffin section: hematoxylin-eosin. magn. 700 x

Fig. 2. External capsule: pigment contained in the astrocytes, the macrophages and diffusely scattered throughout white matter. Paraffin-section. Hematoxylin-eosin. magn. 600 x

The peculiar feature concerns the pigment noticed in our case. A similar pigment was observed by JERVIS in his first case and by DIEZEL (1957) in all