OPHTHALMOLOGICAL FINDINGS IN INFANTILE TYPE OF NEURONAL CEROID-LIPOFUSCINOSIS

CHRISTINA RAITTA, PIRKKO SANTAVUORI
Eye Hospital and Children's Hospital, University of Helsinki, Finland

A new entity which belongs to the neuronal ceroid-lipofuscinoses has been delineated. The essential ophthalmologic features were absence of the ERG and progressive tapetoretinal degeneration leading to blindness at the age of 2 years. Electroretinography and fluoresceinangiography are of major importance for the diagnosis. The primary site of the retinal lesion seems to be the receptors and the pigment epithelium.

Ocular symptoms are principal features of the amaurotic idiocies to which the so-called neuronal ceroid-lipofuscinoses belong. The terminology neuronal ceroid-lipofuscinosis was introduced by Zeman and Dyken in 1969. The disease belongs definitely to the group of inborn errors of metabolism.

The group has been divided into two types: the Bielschowsky-Jansky form, in which visual disability has been considered of minor importance, since it develops when the patients are already severely debilitated, and the Spielmeyer-Sjögren type, which has a chronic course. In this second type macular changes lead to visual disability of importance for the patient because of the protracted course of the disease. These patients are often primarily seen by the ophthalmologist.

Santavuori et al. (1973) have delineated a new entity which belongs to the neuronal ceroid-lipofuscinoses with early onset, exceptional clinical features, and a rapid course leading to a decerebrated stage. Brain tissue shows neuronal obstruction, macrophages in cerebral cortex, astrocytic hyperplasia, and hypertrophia. Remaining neurons and macrophages contain substance with the histochemical features of lipofuscin (Haltia et al. 1973). The new form has been called infantile type of neuronal ceroid-lipofuscinosis. The clinical features of the disease have been presented by Santavuori at this meeting.

A total of 34 children of the series of 52 diagnosed cases have been examined neuroophthalmologically. Examination included evaluation of vision, squint, pupillary reactions and ophthalmoscopy, as well as fluoresceinangiography and electroretinography in general anaesthesia. The age of the children ranged from 12 months to 7 years. Visual impairment, and frequently squint, was observed as early as at 12 months. All children had clearly decreased vision at the age of 18 months. At the age of 2-2.5 years the patients had no light perception, the direct pupillary reaction was absent, and eye movements became incoordinated. In the early stage the fundus was considered ophthalmoscopically normal in 3 cases. However, all had changes in the fluoresceinangiogram and an abolished ERG.

The fundus was hypopigmentated, choroidal vessels were clearly visible. In one boy of 12 months, who belonged to an affected family, a normal ophthalmoscopic picture but definitely pathologic

Proc. 4th Int. Congr. Neurogenet. Neuroophthalmol. (1973)
Acta Genet. Med. Gemellol. (*Roma*), 23: 193-195

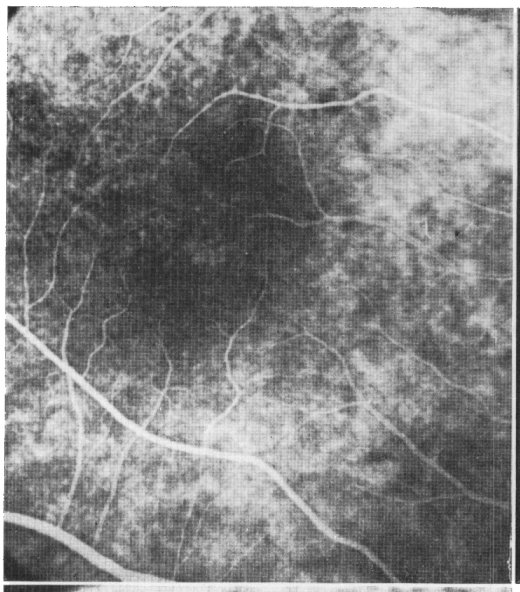

Fig .1

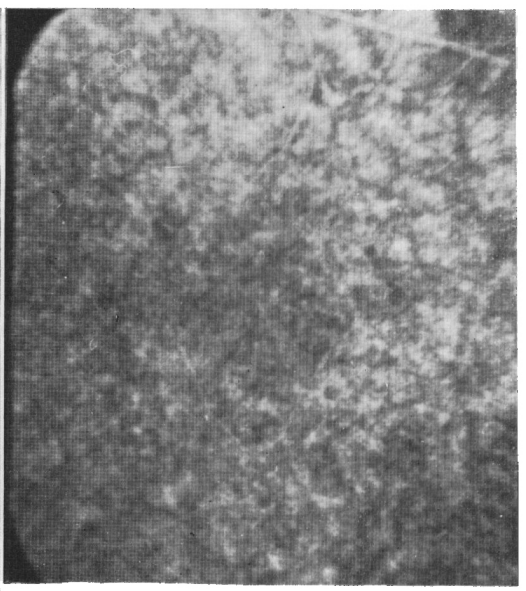

Fig. 2

Fig. 3

Fig. 1. Fluoresceinangiogram of a 12 months old boy belonging to an affected family.
Ophthalmoscopy was considered normal but the flluoresceinangiogram showed already a pathologic pattern of the pigment. ERG extinguished.

Fig. 2. Fluoresceinangiogram of an affected boy of 3 years, brother of the former boy.
Dystrophic pattern, macula hardly recognizable. Pathologic pigment pattern. ERG extinguished.

Fig. 3. Late stage of the disease. Severe retinal dystrophy, especially of the macula. Optic atrophy.

pigment pattern of the fluoresceinangiogram and an extinguished ERG (Fig. 1) indicated that ophthalmologic findings are present even before neurologic clinical signs. Later the retina becomes progressively dystrophic, it loses its lustre, macular changes are prominent, and an ascending type of optic atrophy occurs (Fig. 2). At this stage the child is practically blind.

Electroretinography was performed in all children examined.

Copenhaver and Goodman (1960) studied the ERG in the Bielschowsky-Jansky and Spielmeyer-Sjögren type of neuronal ceroid-lipofuscinosis, and found it usually absent or mark-

https://doi.org/10.1017/S1120962300023751 Published online by Cambridge University Press

edly reduced as a sign of involvement of the outer retinal layers. Normally the ERG is known to undergo distinct changes during different stages of embryonic and postnatal development. The essential features in human ERG during the first year of life are the small potientials that can be recorded. For this reason it seemed essential to study especially those children without recordable ocular symptoms who belong to affected families with manifest disease. The ERG was found abolished in all children, even in those with normal ophthalmoscopical finding, and even in one boy without neurologic symptoms.

REFERENCES

Copenhaver R.M., Goodman G. 1960. The electroretinogram in infantile, late infantile and juvenile amaurotic familial idiocy. Arch. Ophthalmol., 63: 203-209.

Haltia M., Rapola J., Santavuori P., Keränen A. 1973. Neuronal ceroid-lipofuscinosis of early onset. Part 2. Morphological and biochemical studies. J. Neurol. Sci., 18: 269-285.

Raitta C., Santavuori P. 1973. Ophthalmological findings in neuronal ceroid-lipofuscinosis of early onset. Acta Ophthalmol., 51: 755-763.

Santavuori P., Haltia M., Rapola J., Raitta C. 1973. Infantile type of so-called neuronal ceroid-lipofuscinosis. Part 1. A clinical study of 15 patients. J. Neurol. Sci., 18: 257-267.

Zeman W., Dyken P. 1969. Neuronal ceroid lipofuscinosis (Batten's disease). Relationship to amaurotic family idiocy. Pediatrics, 44: 570-584.

Christina Raitta, M.D., Helsinki University Eye Hospital, Haartmaninkatu 4c, 00260 Helsinki 26, Finland.