CASE NOTES

INFANTILE CHOROIDO-RETINAL DEGENERATION WITH CEREBRAL SYMPTOMS*

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ALTHOUGH many cases of choroido-retinal degeneration occurring in children can now be shown to be due to toxoplasmosis, there remains a group which might appear to be due to the same cause but which does not give the necessary positive serum reaction. Such a case is described below.

Case Report

History.—A male infant aged 11 weeks was first brought to the Glasgow Eye Infirmary on March 16, 1948. His mother stated that he seemed unable to follow objects with his eyes. This was her second child and he had weighed 9 lb. at birth. The child's pupils reacted to light but there was very little attempt to follow a light.

Ophthalmoscopic Examination.—This was carried out under general anaesthesia on March 20, 1948, when the appearances in the right eye were as shown in the Figure. Large areas of choroido-retinal degeneration were scattered over both fundi, the macular area being severely involved. The degenerative areas were gyrate in shape and there were irregular patches of pigment scattered throughout the fundi. Both optic disks were somewhat pale and the retinal arteries narrow. The Wassermann reaction was negative.

Later Developments.—On January 15, 1949, no substantial change was noted. The child was x-rayed on this occasion, and no intracranial calcification was found, but the skull was found to be large with a tendency to hydrocephalus. He had had his first tooth at 6 months, and sat up at 7 months.

At the age of one year he was admitted to the Craigmillar Blind School and at about 21 months he had had a short convulsion lasting about one minute. He began to walk at about 2 years. He was discharged from the school as uneducable in April, 1950, at the age of about 2 years and 3 months.

FIGURE — Right fundus.

* Received for publication June 11, 1952.

He had no further convulsions until May 9, 1950, when he had a much longer one accompanied by defaecation, and followed by vomiting. He was then admitted to the Royal Hospital for Sick Children, Glasgow, under the care of Dr. J. H. Hutchison. An x ray of the skull again showed no calcifications, but a pneumoencephalogram showed definite internal hydrocephalus. Cytoplasm modifying antibody (dye) tests on both mother and child for toxoplasmosis were negative. The toxoplasmin test and Mantoux test were also negative. In Dr. Hutchison's opinion, the child certainly did not have toxoplasmosis, and was mentally a low-grade defective.

During 1950, the child had chickenpox and measles. As he grew older it became increasingly obvious that he was mentally retarded. When he was about 3 years of age, he would lie on his back and bang his head on the floor. By this time, however, he was walking and talking a little, and, although his sight was obviously very defective, he could grasp objects held out to him. In view of the gross changes in the fundi, this was surprising, and the eyes were becoming myopic.

When last seen on February 9, 1952, the fundus changes were substantially the same.

Mother's Obstetrical History.—The mother had had in all three pregnancies. There had been no miscarriages.

(1) The first child (by a former marriage) had died from diphtheria at the age of 3 years.

(2) When the second child (the present patient)* was born, the pregnancy was normal and the child was recorded as normal. The antenatal history (from the Glasgow Corporation Clinic) showed slight albuminuria; Wassermann reaction negative; Rh positive.

The only other relevant item of information was the mother's statement that when 3 months pregnant she had had "influenza" which caused her to have a very itchy skin. It is thought possible that she had suffered in fact from German measles, though this cannot now be established.

(3) The third pregnancy† resulted in a stillbirth. The mother was admitted to hospital on July 12, 1950, with severe pre-eclamptic toxæmia when 7 months pregnant. She was also very anaemic (haemoglobin 35 per cent., red blood count 1,800,000). The foetus died and she was finally delivered of a macerated stillborn child on August 8, 1951.

Family History.—In view of the large round areas of choroidal atrophy it was thought that the case might be one of gyrate atrophy of the choroid, and seven of the child's relatives were therefore examined. These comprised both maternal grand-parents, three paternal uncles, one paternal aunt, and one maternal aunt. All were found to have normal fundi. In addition, the paternal grandfather's records were consulted at the Glasgow Eye Infirmary, and it was found that he had suffered from senile cataract, successfully operated upon by Dr. Pendleton White. The child's maternal grandfather stated that he had had a cousin who was born blind, but she had died many years ago in an institution and it was not possible to find out the cause of this blindness. The child's parents were not related before marriage.

Discussion

Although the circular areas of choroidal atrophy suggested a diagnosis of gyrate atrophy, the early onset, central rather than peripheral distribution, and lack of a familial tendency, all made such a possibility appear unlikely.

The mental deficiency, the tendency towards hydrocephalus, and the

* I am indebted to Dr. R. G. H. Cunningham, medical superintendent of the Glasgow Eastern District Hospital, for particulars of the mother's confinement in 1947, and of the present patient's birth.

† I am indebted to Dr. W. Gordon Miller, obstetrical registrar at Robroyston Hospital, Glasgow, for these particulars.

convulsions, suggested a diagnosis of toxoplasmosis, but x-ray examinations, both at the Glasgow Eye Infirmary and at the Royal Hospital for Sick Children, Glasgow, revealed no intracranial calcification. In addition, the serological tests for toxoplasmosis were negative in both mother and child.

Sabin and Feldman (1949), in a review of 43 children with choroidal atrophy and mental deficiency, state that, of 23 children with positive serological reactions for toxoplasmosis, 90 per cent. showed grossly perceptible cerebral calcification in x-ray photography of the skull, and that, in a group of twenty children with choroidal atrophy and negative serological tests for toxoplasmosis, the incidence of cerebral calcification was only 5 per cent. The presence of cerebral calcification, therefore, is suggestive of toxoplasmosis, but its absence does not preclude a diagnosis of toxoplasmosis although it makes it less likely. This is borne out in the present case.

Sabin and Feldman add that, in cases of infantile chorioretinopathy without cerebral calcification or evidence of toxoplasmosis, the available data suggest defective development rather than destructive, necrotic lesions as the more probable pathological basis.

Richards (1951) reported a case of microcephaly with chorioretinopathy, cerebral calcification, and internal hydrocephalus, but with negative serological reactions. In this case, the left fundus showed a localized macular patch of chorioretinopathy and peripheral granular pigmentation. The right fundus (not depicted in the article) showed generalized retinal atrophy and some peripheral granular pigmentation with a normal macula. The picture of the left fundus does not resemble that shown in the present case, since in the former, the degeneration is practically localized to the macula, whereas in the latter the degeneration is very widespread, involving the macula in addition to other areas. In each case a pneumo-encephalogram showed internal hydrocephalus, but in Richards's case marked atrophy of the right cerebral hemisphere and calcification in the left parietal area related to the left lateral ventricle were also demonstrated. In Richards's case the cytoplasm modifying antibody tests and complement-fixation tests were all negative in the child but positive in the mother. In Sabin and Feldman's cases, as in that here reported, the maternal titres were all negative or of insignificant level.

The aetiology of the present case therefore remains obscure, but the possibility that the mother suffered from German measles during her pregnancy may have some bearing on the child's condition. In addition, the mother appears to have had symptoms of pre-eclampsia, moderate in the pregnancy concerned, but much more marked in a subsequent pregnancy which resulted in a stillbirth.

Summary

A case of extensive choroido-retinal degeneration occurring in infancy and associated with mental deficiency and convulsions is described. Serological

tests for toxoplasmosis in mother and child were negative and the aetiology is obscure. In the third month of pregnancy the mother suffered from a febrile illness which may have been German measles. She also suffered from albuminuria in the pregnancy concerned, and from severe pre-eclamptic toxæmia in a subsequent pregnancy ending in a stillbirth.

I am indebted to Dr. J. H. Hutchison, Physician, Royal Hospital for Sick Children, Glasgow, and to Drs R. G. H. Cunningham and M. A. Foulis, Medical Superintendents of the Glasgow Eastern District Hospital, and the Robroyston Hospital, Glasgow, respectively, for permission to publish their notes on this case. My thanks are also due to Mr. G. Donald who did the fundus painting.

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