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INTRA-OCULAR PHAKOMATA—A REPORT OF THREE CASES*

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IN 1932 van der Hoeve grouped the syndromes of Bourneville, of von Hippel and Lindau, and of von Recklinghausen under the title of the "phakomatoses." Later the syndrome of Sturge-Weber was added, making a fourth.

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Received for publication, April 24, 1948.

He called the tumefactions found in these diseases "phakomata," from "phakos," the Greek name for mother spot. He described by the word "phakos," a spot, congenital in origin, often hereditary and familial, and which can be found in different parts of the body. It can be present at birth or appear later on, can vary in size, enlarge by proliferation of any part of the tissue, grow to real blastomata and even turn to malignancy. A phakos may be present in any part of the human body.

All the phakomatoses have an hereditary character, although direct evidence of this may not be discovered in many families. The full syndromes are often not seen in all members of a family and mental changes may be completely absent. The discovery of retinal phakomata may be most important in clinching an otherwise doubtful diagnosis. The three patients described below have very obvious stigmata, but all show interesting ocular features that have been considered worthy of record.

Two are cases of phakomata-Bourneville (tuberosc sclerosis, epiloia) and one of phakomata-von Recklinghausen (neuro-fibromatosis). The patients were low grade idiots with epilepsy. Each showed cutaneous and retinal spots (phakomata). Intracranial lesions must have been present in addition.

In the family histories no similar conditions are recorded, but the relatives could not be investigated for incomplete manifestations of the syndromes.

Case Reports

CASE NO. 1. E. H. K. Female. Born 1908, died 1936. In 1932 when admitted to mental hospital she was stubborn and resistive. Her physique was poor and she had a left-sided hemiplegia. Adenoma sebaceum was present on the face over the butterfly area and chin. There were some fine spider naevi among the nodules. The pupils were noted as equal and reacting to light and a routine photograph showed left convergent strabismus (Fig. 1). Major epileptic fits numbered about a dozen per annum.

In January, 1933, both pupils were noted as slightly enlarged, the right more than the left, and reacting sluggishly to light.

In September, 1933, the patient's vision appeared to have deteriorated. Early bilateral papilloedema was found with the retinal spot (Fig. 3). A month later a right ptosis and weakness of right internal rectus muscle developed (Fig. 2).

During 1934 the patient became stuporose, incoherent in speech and appeared oblivious to her surroundings. At times she cried out with pains in the head.

FIG. 1.

Case No. 1 showing adenoma sebaceum, left internal strabismus.

FIG. 2.

Case No. 1 showing adenoma sebaceum with right ptosis and weakness of right internal rectus muscle.

During 1935 the pupils became dilated with no reaction to light. Both discs were dead white with very small retinal vessels (secondary optic atrophy). Coarse nystagmus was present. The ptosis and strabismus persisted. During the year she showed signs of further increase in intracranial pressure, the fits became more frequent and she lived a "vegetable existence" until death in December, 1936.

Post-mortem showed numerous tuberous masses in the cerebral hemispheres, particularly in the temporal lobes. There was a large spongioblastoma within the third ventricle with advanced internal hydrocephalus.

Fig. 3 shows the right fundus when papilloedema was present. Just above and temporal to the disc, surrounded by the branching superior temporal artery, there is a sharply defined, circular, flat mass within the retina. Its size is slightly larger than the optic disc. Its colour is chalky white with streaks of yellow and its surface is rough and coarsely pitted. It is surrounded by a broad slate-grey matte ring that gradually fades into the surrounding retina with slight radial extension towards the optic disc. It is a typical retinal phakos as found in Bourneville's disease.

The case is interesting in showing the syndrome of epiloia with retinal phakos, and ventricular tumour causing internal hydrocephalus and raised intracranial pressure leading to ocular palsy and secondary optic atrophy.

CASE No. 2. F. S. Female. Born 1900, still alive aged 48 years. She is fifth in a mixed family of six; the others are said to be normal. She is unstable, destructive, impetuous and resistive unless managed very quietly and patiently. She is unable to do any of the routine mental tests. Epileptic fits number six to twenty per annum.

She shows very pronounced adenoma sebaceum of the face, particularly on the naso-labial folds and chin. There are some smaller nodules on the upper lip and fewer of varying size on the forehead (Fig. 4). Fine spider naevi are present in association with the nodules.

Ocular examination. No abnormality has been detected in the right eye. Except for the phakomata, the left fundus (Fig. 5) is normal. Below and temporalwards from the left disc, surrounded by branches of the inferior temporal vein, are two chalky-white masses close together within the retina. Each is semi-circular, in size about one quarter of a disc diameter. Their surfaces are flat and finely rough. They are surrounded by a slate-grey area that gradually fades into the surrounding retina. They are retinal phakomata of Bourneville's disease. No change has been noted for many years.

FIG. 3.

Case No. 1. Tuberose sclerosis—retinal phakos, papilloedema.

FIG. 5.

Case No. 2. Tuberose sclerosis—retinal phakomata.

FIG. 7.

Case No. 3. Neurofibromatosis of choroid, circulatory disturbances around macula, secondary optic atrophy.

FIG. 4.

Case No. 2 showing extensive adenoma sebaceum of case.

FIG. 6.

Case No. 3 showing small pigmented spots on forehead. Subcutaneous neurofibromata are not seen in photograph.

The patient is interesting in showing the above features and for having lived so long despite her handicaps. For several years she has had, in addition, active pulmonary tuberculosis. This has slowly progressed to become bilateral, but has elicited a very good fibrotic response and her general physique is good. There is no evidence of raised intracranial pressure.

CASE NO. 3. H. C. Male. Born 1898, still alive aged 50 years (Fig. 6). His mental deficiency was noted when he was aged 5 years. To his family's knowledge he is the only mental case for four generations.

On admission in 1931, he was found to be a low grade imbecile who spoke little and replied mostly by signs. At first he had infrequent attacks of petit mal, but over several years these increased to fifty to eighty per annum with some grand mal attacks.

His skin shows several patches of brown pigmentation, particularly on the abdomen, with a few on the face. Multiple subcutaneous fibromata are scattered over the entire body. Numerous venous varicosities are present in each groin and in other sites near the fibromata. The third or fourth lumbar spine appears deficient (possible spina bifida occulta) and both knee and ankle reflexes are absent. His general condition has changed little since admission.

Ocular examination. Tension by palpation appears normal. Both optic discs show "secondary" optic atrophy. Their outlines are finely irregular and both are of a pale yellow colour. Otherwise the right fundus appears normal. The left fundus (Fig. 7) shows a circumscribed, slightly raised, pale area close to the superior temporal vessels. It is irregularly quadrilateral, appearing somewhat larger than the optic disc. Over it the usual fundus redness is replaced by a pallor, and some choroidal vessels can be seen in its depth. In places it is bordered by a little fine pigment and a clear rim that quickly fades into the surrounding fundus. It has the appearance of an almost flat tumour within the choroid, slightly raising the overlying retina. At the macula there are some fine irregular pigment deposits and fine exudates suggestive of circulatory disturbance. The phakos is probably a neurofibroma of the choroid. The bilateral secondary optic atrophy is probably due to similar tumours associated with the optic nerves. The patient cannot be X-rayed owing to his mental state.

Summary

Three cases of intra-ocular phakomata are recorded. The appearances of the retinal phakomata in the two patients with Bourneville's disease (tuberous sclerosis) are so characteristic that

diagnosis could be made if these were the only stigmata found. The diagnosis of the probable choroidal neurofibroma depends on the presence of very numerous, widely spread neurofibromata readily identified elsewhere, with bilateral secondary optic atrophy, epilepsy and amentia. On appearances alone its diagnosis could not be differentiated from other choroidal tumours.

Acknowledgments.—These investigations were conducted from the Institute of Ophthalmology, University of London. My thanks are due to the Dean (Mr. R. C. Davenport) and the Academic Board for the facilities provided. The patients were examined at Leavesden Hospital. I am most appreciative of the kindness and co-operation given me by the physician-superintendent, Dr. J. K. Watkin, and of the clinical help given me by Dr. W. M. McGrath. I have drawn freely on the clinical records and museum material of the hospital.

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ANNOTATIONS

Results and causes

To those who insist on putting first things first our title will be of the nature of an hypallage—the cart before the horse. In ophthalmology, especially in hospital practice, it is astonishing to what a variety of causes the O.P. is apt, in moments of expansion, to attribute the condition for which he or she seeks advice. Mothers often opine that a child's squint has resulted from attempts at copying some other squinter, either at home or among friends; and the same is also said of chorea. We recall an elderly male out-patient with a tarsal cyst who said it was hereditary. Perhaps it was, we did not feel called upon to question his statement.

There is really no knowing to what a patient will ascribe his condition. Tristram Shandy thought that the "asthma," from which he suffered, was due to skating against the wind in Flanders; but we do not believe that any author of a text-book on medicine has ever seriously included it among the causes of asthma. Sterne was, of course, a consumptive, and gives a very odd cause for a fit of laughter which brought on an attack of haemoptysis. It is not suitable for inclusion here, but will be found in the original (Tristram Shandy, Vol. VIII, Chapter 6, of the collected edition of Sterne's