CUTANEOUS NAEVUS WITH BUPHTHALMOS AND EPILEPSY: CASE REPORT.

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IN 1879 Sturge gave the first complete account of a rare syndrome comprising the co-existence of naevus flammeus of the skin, glaucoma or buphthalmos and epilepsy.

The naevus flammeus appears characteristically over one side of the face within the distribution of the divisions of the trigeminal nerve, occasionally bilaterally and rarely extending over segmental areas of the body as in the case to be described below. The buphthalmos or glaucoma occurs on the same side as the facial naevus. Histologically in several cases the eye has shown a proliferation of the choroidal blood vessels or a venous angioma. Epilepsy, hemianopia, hemiplegia and feeble-mindedness are all described in cases conforming to the syndrome.

Within the cranial cavity two types of lesion have been described, viz.: (*a*) a venous cavernous angioma of the meninges and brain, sometimes penetrating as deeply as the ventricular system; or (*b*) telangiectasis of the pia arachnoid with shrinkage and sclerosis of the underlying cortex and deposits of lime-salts in the second and third cortical layers, but not in the pial vessels (Krabbe). Parkes Weber (1922) added a fourth sign to the syndrome—the characteristic X-ray appearance in skiagraphs of the skull. Skiagraphs may show sinuous lines of calcification with double contour, usually situated in the occipital region, indicating the position of calcified cerebral gyri and sulci. These appearances are not constant and occur characteristically with the second type of intracranial lesion.

Brouwer, van der Hoeve and Mahoney (1937) relate the syndrome to other heredo-familial diseases exhibiting ocular, nervous and cutaneous symptoms which they term collectively phakomatoses, from the Greek word φακός, meaning "motherspot" or "mole." The other diseases related by these workers in this way are tuberose sclerosis and neurofibromatosis.

Rönne (1937) collected some two hundred cases of the syndrome from the literature. Some authors include within the syndrome cases of "naevoid

amentia," although many of the cases, like ours, have not been "aments." Not all of the cases show the full syndrome; in about a third of the cases ocular lesions are present. Nussey and Miller (1939) recently published a case report, and their paper gives a very fine bibliography which will not be repeated here.

Case History.

A soldier, aged 26, was admitted on December 2, 1940, because of repeated fits while with his unit. His father, a gamekeeper, was accidentally shot. His mother, whom he had not seen for five years, was at that time alive and well. One half-sister, whom he had not seen for ten years, was also well. No member of the family was known to have birthmarks or fits. The patient, whose naevi had been present at birth and had not changed in size or distribution, had had fits and been blind in the right eye as long as he could remember. At school he had managed to learn fairly adequately but was always rather backward. His work record was not good, but he had managed to keep a job as "runner with a firm of coal and oil merchants." He had never felt fit for heavy work. At one time he had done some lorry-driving and had never had a fit during this time. He had managed to get into the Army by suppressing evidence of his fits. The fits, which were very frequent in childhood, had been less so during the last five years.

Description of fit.—The fit is preceded by a visual aura in which he sees zigzags and stars in the left half of the visual field. These are moving and of various colours. The aura lasts for about five minutes and is associated with a blackness, so that he can see only parts of people. He describes this as seeing people "moving in bits." The aura always proceeds to a fit and the patient sits or lies down; then he knows nothing more. He experiences no paraesthesiae or olfactory or auditory phenomena. On regaining consciousness he cannot speak for about 15 minutes and the left arm and leg remain useless for about half an hour. The attacks are associated with severe headache and vomiting. He has no memory of having had urinary incontinence during the attacks. Luminal appears to have made very little difference to the frequency of the fits.

Physical examination.—On examination the patient, who is a right-handed man, presents a slightly boyish appearance for his years, but shows normal secondary sex characters. He gives a clear history of his life and is not regarded as mentally defective, though his intelligence is certainly low; the Intelligence Quotient was 72. His behaviour and general social adaptation in the hospital were excellent. The right side of the face is larger than the left, the nose pointing to the left, and the growth of hair on the right side is slightly less thick. At rest the right eye deviates inwards. The pupil is almost inactive to light, but reacts to accommodation (Mr. A. J. Cameron, F.R.C.S., Ophthalmologist to the Hospital, has kindly reported on the eyes).

Report on eyes (Mr. Cameron): "Right eye, buphthalmos. Right cornea several millimetres wider than left. Deep anterior chamber. Marked optic atrophy and deep cupping of disc. Hence blindness resulting from congenital glaucoma. Corneal nebulae. Limitation of movement due to amblyopia. No retinal vascular abnormalities beyond some venous congestion noted. Left eye, no abnormality except misshapen disc."

The naevus (Figs. 1, 2), which is of a reddish colour with a blue tinge, involves the whole of the right trigeminal area, including the right side of the soft palate and tongue. Over the forehead and malar region the skin is darker red and nodular. The right side of the neck and forequarter, the flexor and radial aspects of the right arm and palm of the hand are also involved. On the left side the third division of the trigeminal area, and the ulnar aspect of the left arm and hand, show the same naevoid condition.

Apart from the right eye, there is no abnormality in the cranial nerves or in the rest of the central nervous system. The reflexes are brisk and equal, the plantar

responses flexor, and the power in the limbs equal and normal. There is no ataxia and no sensory loss.

Examination of the C.S.F. revealed normal pressure and no qualitative abnormality. Radiographs showed no abnormality and no calcification. Air-replacement encephalography was performed by the lumbar route and 80 c.c. of air introduced. No abnormality of the ventricular system was seen, except that the right lateral ventricle was slow in filling. After the head was placed with this ventricle uppermost for ten minutes a certain amount of air entered, but the anterior horn still did not fill. An electro-encephalogram was taken by one of us (D. H.) and revealed no abnormality. Normal rhythms were present. This is in conformity with the view that, apart from idiopathic epilepsy, E.E.G. abnormality is found only where there is an active histopathological lesion in progress.

The patient had no fits in hospital, and after invaliding from the Army returned to his old occupation as "runner."

FIG. 1.

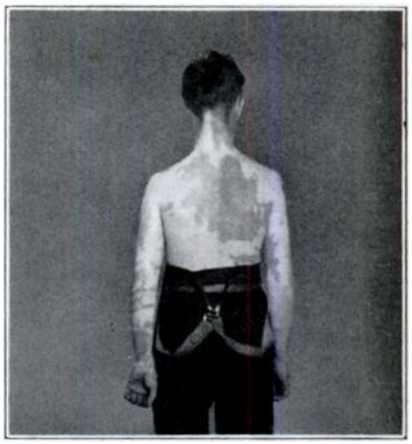

FIG. 2.

DISCUSSION.

In this case the naevus flammeus is unusually widespread in its distribution, involving the right trigeminal area and the trunk and upper limbs, together with the third division of the left trigeminal nerve. The buphthalmos of the right eye is possibly due to some vascular abnormality of the ciliary body or choroid, although none can be demonstrated clinically. The contralateral fits which the patient describes resemble migrainous attacks in that they are preceded by teichopsia. The loss of consciousness and residual transient paresis of the left arm and leg, however, render it probable that he suffers from a type of Jacksonian fit, although no fit was actually witnessed in hospital. The nature of the intracranial lesion responsible for these contralateral fits can only be surmised. The absence of calcification in the skiagram and the slow filling of the right ventricle with air on ventriculography suggest the presence of a venous angioma of the meninges and brain rather than the type

of lesion described by Krabbe. In our patient no heredo-familial incidence of the syndrome in whole or part can be ascertained.

SUMMARY.

A report of a case of Sturge's syndrome is given showing diffuse naevus flammeus, buphthalmos and Jacksonian fits of the contralateral side of the body. The patient was of low mentality with an intelligence quotient of 72, but by no means mentally defective. Radiographs of the brain showed no calcification of the cerebral sulci. Results of air-replacement and electro-encephalography are described.

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