DIFFUSE NEUROFIBROMATOSIS WITH PROPTOSIS

BY

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DIFFUSE neurofibromatosis in which the eyelids, orbit and surrounding parts are involved, is a well-recognized picture, and a good many examples of it have been fully reported in this country. At different times I have had four cases under my care and the last one being a good example with, as is usual, several points of interest, it seems worth while to report it in detail, and at the same time to refer, with regard to particular points, to the other three cases.

Clinical Details

H. M., aged 33 years, male, was sent to me at St. Bartholomew's Hospital by Dr. A. C. Roxburgh to see if it were possible to improve his condition. I admitted him to my ward on June 18, 1930. The patient's tale was that his right eye was quite normal until the age of six months when he fell downstairs, and following this accident the eye gradually became pushed forward. At the age of seven he attended the Royal Victoria Eye and Ear Hospital, Dublin, and Dr. Werner has been good enough to look up the notes of that date, and has sent me the following particulars:—He was seen in December, 1903, with the history that the right eye had been protruding for three months. At this time there was slight proptosis which was increased by muscular exertion, after which marked pulsation of the globe was felt and a faint bruit was heard over the malar bone. Later the condition got markedly worse, the eye being pushed downwards and forwards. No tumour was felt and there was no engorgement of the retinal veins of this side. The interior of the orbit was exposed from the outer side by partial resection of the outer wall and a few weeks later the carotid artery was tied, as a result of which the proptosis is stated to have been much less and no pulsation was felt on leaving the hospital. He has been to a number of hospitals since then but does not think the eye has changed; he thinks it protrudes more than ever when he is excited and can then feel it throbbing and pulsating. Apparently the chief disadvantage from which he suffers is his inability to obtain work. He states that the first of the nerve tumours of the skin to appear was the one in the left hypochondrium (Fig. 1) and that they have gradually increased in number since. There has been no pain in any of the lumps and indeed they cause him no inconvenience.

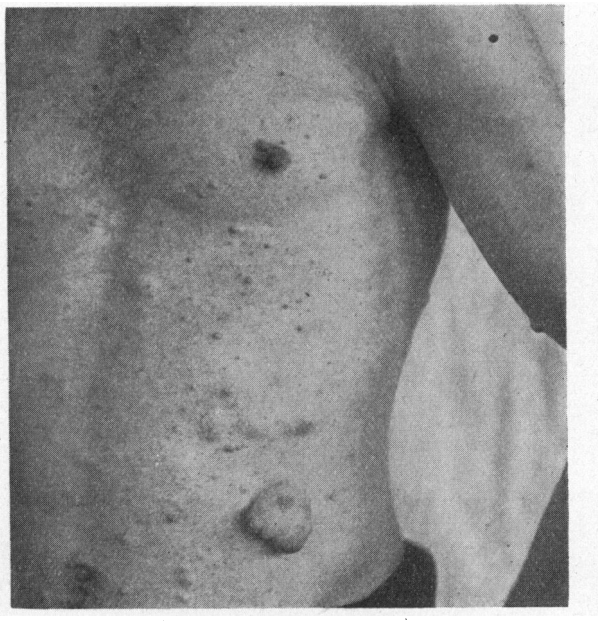

FIG. 1.

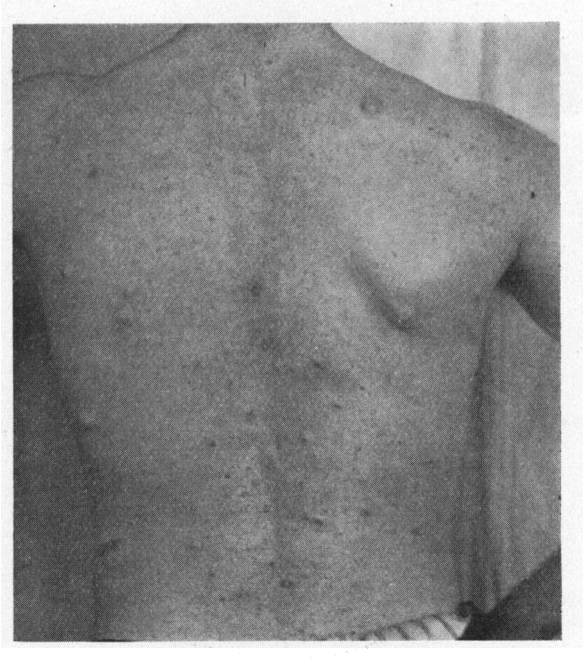

FIG 2.

He has occasional frontal or occipital headaches. He is not aware of any defect of sight, and in spite of the great displacement of the eye has never had diplopia or any other visual disturbance.

Alexis Thomson has pointed out that neurofibromatosis is not infrequently familial and hereditary, but this patient's father and mother, two brothers and two sisters are alive and well, and without any similar condition of the eyes, nor, so far as he knows, of the skin. The distribution, number and arrangement of the nodules is well seen in the accompanying photographs (Figs. 1 and 2). The largest one is on the abdomen and is two inches in diameter. Many of them are only a few millimetres across. The great majority are intra-dermal, those which are subcutaneous are movable in one plane only, suggesting that they are attached to a longitudinally-running structure.

The Skull

Measurements: circumference 61 cms. (average male 52·5), length 20 cms., breadth 16·4 cms. Cephalic index, breadth $\times$ 100 / length = 82, that is, he is brachycephalic (Fig. 3).

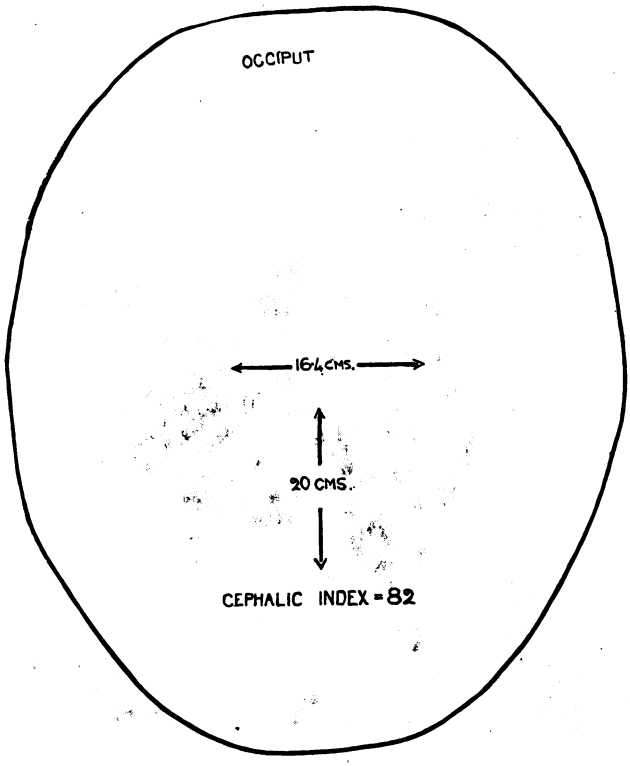

FIG. 3.

X-ray Examination

The right orbital cavity is considerably larger than the left and no details of the posterior orbital walls are seen (Fig. 4) apparently

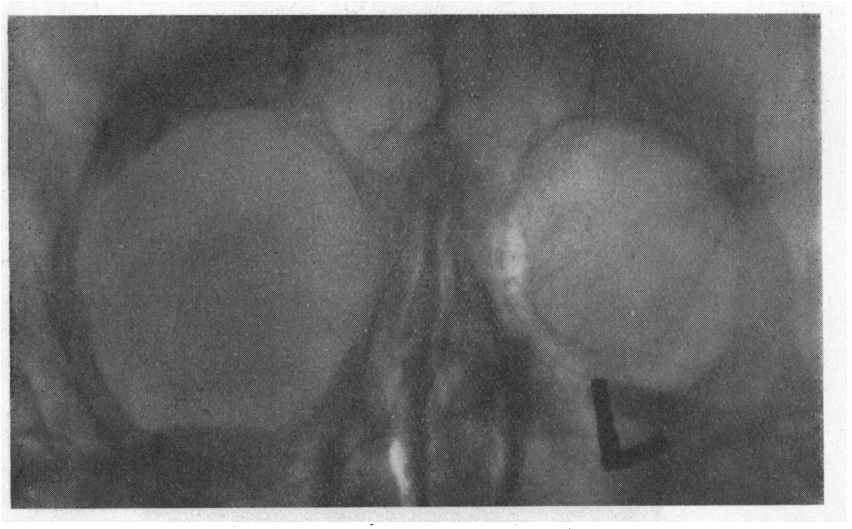

FIG. 4.

as a result of malformation or deficiency, providing therefore a free connection between the orbital and cranial cavity.

The Eyes

Marked forward and downward displacement of the right eye is well seen in the photograph (Fig. 5). It is possible to get one's fingers so far behind the globe as to feel the optic nerve. The lids close well over the eye during sleep, and the movements are but little restricted. On placing the flat of the hand upon the closed lids pulsation is felt; it is not, however, of the expansile type as occurs in aneurysm, nor is the proptosis appreciably reduced by pressure. The fundus is normal, the visual acuity is 6/9 and the field of vision is complete. (Figs. 6 and 7). The left visual acuity is 6/9 also, the visual field too is full and the fundus is healthy. Adjoining the upper edge of the disc in this eye is a patch of medullated nerve fibres. No abnormality was found in the urine, the blood sugar of the capillary blood an hour and a half after a meal was 106 mgs. per 100 c.c.

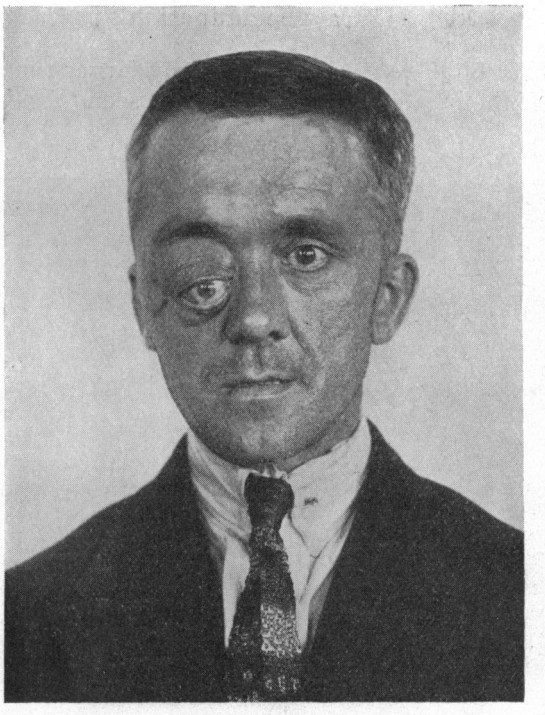

FIG. 5.

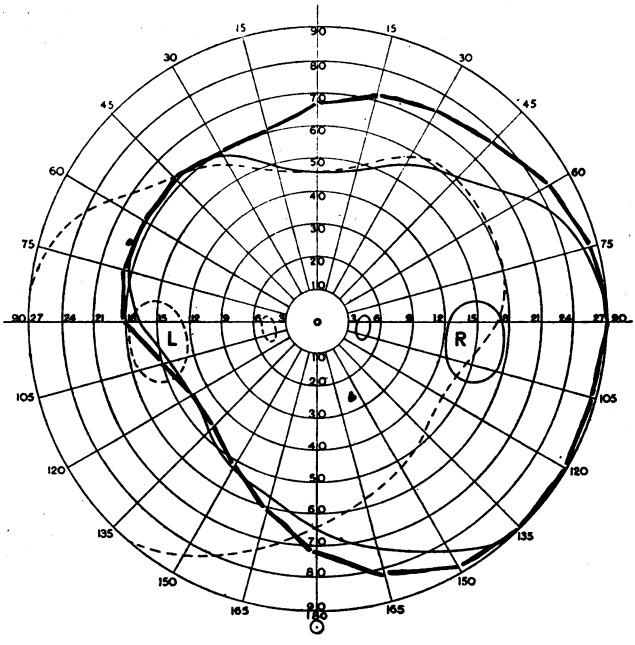

FIG. 6.

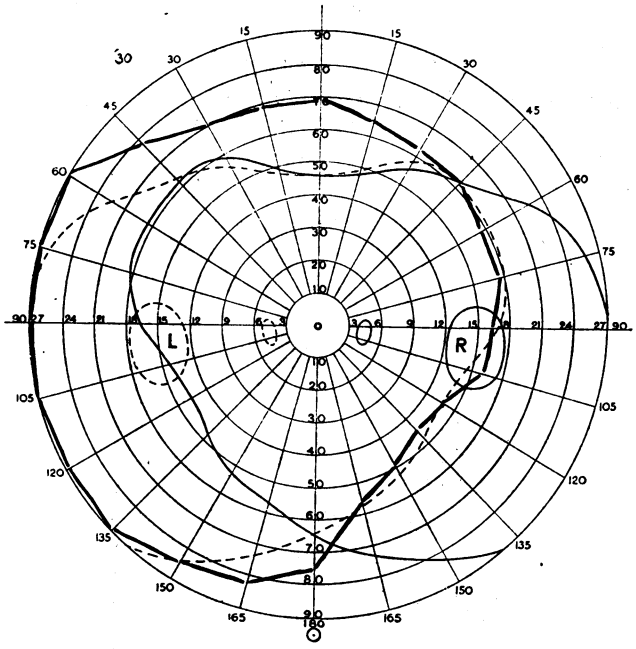

FIG. 7.

The earliest account of collected cases of tumours of nerves was by William Wood (" Observations on Neuroma," *Edin. Med. Chir. Trans.*, Vol. III, 1829) and it is worthy of note that he believed they arose from the connective tissue sheaths of nerves. He quotes a case from Cheselden's " Anatomy of the Human Body," in which the tumour was removed.

A number of other cases were then reported until R. W. Smith's classical work appeared in 1849 (" A Treatise on the Pathology, Diagnosis and Treatment of Neuroma," Dublin, 1849) and of recent date Alexis Thomson's outstanding monograph " On Neuroma and Neurofibromatosis, Edinburgh, 1900 " should be consulted for information on every branch of the subject.

There are several points which are exemplified in these cases to which I should like to draw attention.

The history does not always commence at birth although the condition is probably present then, the skin tumours not infrequently, as in the present case, increase in number at about puberty.

Thomson has shown that many cases of diffuse neurofibromatosis are associated with other tumours of nerves such as plexiform neuroma which is of a precisely similar nature except that it is limited to one nerve or nerve trunk, intracranial tumours, tumours

of the chiasma, etc., but none of these was exemplified in the present case.

The point to which I should like to direct attention is the pulsatile nature of the proptosis, and so far as I can see this phenomenon is not mentioned in any of the very numerous cases which are quoted by Thomson, and yet it has occurred in all the four cases I have seen in which proptosis was present. In the present case it had induced a surgeon many years ago to ligature the carotid artery in the belief that the orbital mass was of a

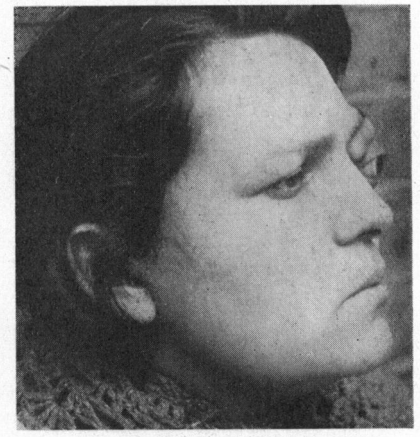

FIG. 8.

vascular nature. Fig. 8 is a picture of a case which Mr. Holmes Spicer has been good enough to send me in which also the exophthalmos was pulsatile.

The pulsation is not of the expansile type that is so characteristic of aneurysm, nor is there the bruit to be heard over the skull.

It will be seen in the skiagram (Fig. 4) that the orbit is much larger than on the opposite side and no bony details are to be seen in the posterior orbital wall. It seems evident, therefore, that the pulsation that is felt is not of direct vascular origin but is transmitted directly to the orbital tissues from the brain.

Rockliffe and Parsons published a case in which the patient, a child of two, died after exenteration of the orbit preceded by ligature of the carotid artery (*Trans. Path. Soc.*, 1903). In their case the whole of the bone of the orbit except the floor and outer wall had disappeared, or I suspect had never developed, so that the growth was in direct continuity with the cranial cavity, a fact which no doubt was responsible for the pulsation.

A second feature which was but slightly marked in the present case but was very marked in one of my other cases is a large brachycephalic skull and especially a fullness or bulging in the temporal fossa. (Ludford Cooper, *Trans. Ophthal. Soc. U.K.*, 1906, XXVI, 136).

In one of my cases this was so marked as to appear as if the squamous bone in particular were being pushed outwards by some tumour within, and in another case although the prominence was less extreme the man had been trephined over the area, perhaps with the expectation of finding a tumour.

In addition to the bony bulging in the temporal fossa, it is common in those cases with large pendulous upper lids due to a plexiform neuroma to find that this condition has extended into the temporal fossa causing a doughy sort of swelling there. (Treacher Collins and Rayner Batten, *Trans. Ophthal. Soc. U.K.*, 1905, XXV, 248, and Snell, *Trans. Ophthal. Soc. U.K.*, 1903, XXIII, 157).

With regard to the skin tumours, if the present patient's observation may be trusted, they first showed themselves about 15 years ago, that is at the age of 18 years, about 12 or 15 years after the development of the proptosis. The presence of the opaque nerve fibres in the left retina may be no more than a fortuitous occurrence; on the other hand it seems likely that they are in some way connected with the abnormality of the other nerves of the body.

I have especially to thank my House Surgeon, Mr. E. G. Recordon, for the details of the clinical history and the general overhaul of the patient, and for obtaining the plaster cast and measurements of the skull.

ANGIOID STREAKS, AND THEIR RELATION TO A FORM OF CENTRAL CHOROIDAL DISEASE

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THIS selection of drawings from Hamblin's collection, together with the family which I report, while of interest in itself, is, I consider, important in the evidence it affords in confirming and consolidating the observations of others on angioid streaks and their relation to a special form of choroidal disease, and in establishing it as a definite disease, with special characteristics of its own, frequently familial in causation.