PULSATING EXOPHTHALMOS DUE TO NEUROFIBROMATOSIS*

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CASES of pulsating exophthalmos, which are seen from time to time, are mostly vascular in origin. Rarely they are due to transmitted cerebral pulsations, as in congenital failure of the orbital roof, erosion of the walls due to inflammatory processes, and traumatic hiatus of the orbital roof. Occasionally they are due to plexiform neurofibromatosis.

von Recklinghausen (1882) first described a combination of melanomata and multiple neurofibromata arising from cutaneous nerve filaments. The complete picture is characterized by tumours of the skin, cutaneous pigment, multiple tumours arising from the sheaths of cranial, spinal, peripheral, and sympathetic nerves, defective development of the central nervous system, mental abnormalities, and buphthalmos. There are various incomplete forms.

Páez Allende (1945) listed the sites of ocular involvement in neurofibromatosis in the order of frequency—the lids, optic nerve, orbit, retina, iris, cornea, tarsal conjunctiva, and bulbar conjunctiva. Cockayne (1933) reported that there is a definite limitation to the male sex in isolated cases of neurofibromatosis. Hine and Wyatt (1928) reported a case of neurofibromatosis of the right orbit. Moore (1931) suggested that pulsatile exophthalmos in neurofibromatosis was caused by dehiscence of the posterior part of the orbit. Lisch (1937) and Wolter and Butler (1963) found that nodules in the iris were always bilateral in these cases. That glaucoma might not occur until adult life has been emphasized by Duke-Elder (1940) and has been illustrated by Meeker's (1936) case.

A case is reported below of von Recklinghausen's disease presenting with multiple cutaneous neurofibromata, *café-au-lait* spots, and plexiform neurofibromata of the upper lid and temporal region involving the right orbit, producing pulsatile exophthalmos, nodules in the iris, and asymmetry of the pupil of the same side.

Case Report

A man aged 20 years was brought to the hospital with a pendulous swelling of the right upper lid which had been progressively growing since birth. He had had defective vision of the right eye for the past five years.

Examination.—The upper lid of the right eye was markedly thickened, flabby, and pendulous, and contained knotted tortuous cords. The right eye could not be opened voluntarily (Fig. 1, opposite). The tumour had spread over the right fronto-temporal region, producing facial asymmetry. The eye, which was of normal size, showed pulsatile exophthalmos of about 5 mm. with downward and inward displacement of the eyeball (Fig. 2, opposite). There was mechanical limitation of movement upwards and on the right side. No bruit was heard.

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FIG. 1.—Plexiform neurofibroma forming a pendulous mass of the right upper lid.

FIG. 2.—Exophthalmos with deviation.

FIG. 3.—X ray of the orbits. Note the diffuse enlargement of the right orbit and the lack of detail of the posterior part of the orbit indicating a free connexion between it and the cranial cavity.

The conjunctiva was thickened and pigmented, the cornea was oedematous, the pupil was horizontally oval, and the iris showed greyish-white nodules on its surface. The vision was reduced to hand movements. The ocular tension was 70 mm. Hg (Schiötz). Gonioscopic study of the angle of the anterior chamber was not done and the fundus view was hazy because of lenticular opacity.

The left eye was normal with full vision and the fundus was normal. The field of vision in that eye was full. No iris nodules were seen.

The general examination showed multiple cutaneous nodules and *café-au-lait* spots. X-ray examination of both orbits showed a diffuse enlargement of the right orbit with lack of detail in the posterior part (Fig. 3).

Summary

This case is interesting for the following reasons:

1. The swelling of the right upper lid had grown more rapidly for the last five years showing that these tumours become activated during puberty.
2. The fact that no other member of the family had this disease is in accordance with Cockayne's finding that isolated cases are limited to the male sex.

3. Pulsatile exophthalmos is due to posterior orbital encephalocele caused by dehiscence of the posterior part of the orbit.
4. The asymmetry of the pupil is due to sympathetic involvement.
5. Usually bilateral iris nodules are reported in cases of von Recklinghausen's disease, but in our case there were iris nodules in the affected eye only.
6. Buphthalmos is commonly associated with this disease; it can occur as secondary glaucoma in the adult at a later date. In this case there was secondary glaucoma and not buphthalmos. The secondary glaucoma may be due to peripheral anterior synechiae, developmental anomalies of the angle of the anterior chamber, or to pressure over the venae vorticosae.

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