PULSATING EXOPHTHALMOS IN VON RECKLINGHAUSEN'S DISEASE

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Unilateral pulsating exophthalmos is often due to an arteriovenous communication between the internal carotid artery and the cavernous sinus, or to an arteriovenous aneurysm in the orbit between the ophthalmic arteries and veins. Occasionally a true aneurysm of the ophthalmic artery may cause exophthalmos. Trauma to the head is a common aetiological factor, but the condition may arise spontaneously. It is not so well known that pulsating exophthalmos can occur in some cases of von Recklinghausen's disease. The underlying cause in these cases is a defect in the bony wall of the orbit which permits the direct transmission of the pulsations from the brain to the globe of the eye. In addition the intracranial contents encroach upon the orbit and give rise to a varying degree of exophthalmos. Examples of this condition have been described by R. F. Moore (1931), LeWald (1933), Wheeler (1936), A. E. Moore (1936), Pfeiffer (1943), Friedman (1944), and Peyton and Simmons (1946). Petit-Dutaillis, Feld, and Taptas (1949) were able to find only 37 references to this condition in the international literature; they added two cases of their own. Such a small number of reported cases is surprising when the voluminous literature on von Recklinghausen's disease is considered. This would suggest that the condition is either rare or that, as pointed out by Peyton and Simmons (1946), the lesion is recognized only by those familiar with the syndrome.

The cutaneous manifestations of von Recklinghausen's disease are known to be only one aspect of a congenital defect which may disturb normal development in many parts of the body. In particular, abnormalities of the skeletal system are now well known and have been reviewed by Holt and Wright (1948). The changes which may be found in the bones of the skull in this disease have been discussed by Rosendal (1938). Osseous lesions, if looked for in von Recklinghausen's disease, are common. Crowe, Schull, and Neel (1956) have expressed the view that it is only rarely a patient

does not show some skeletal involvement. Defects in the wall of the orbit appear to be one of the osseous manifestations of this disease and perhaps they are more common than is at present recognized.

We have seen in the space of a few months two patients with von Recklinghausen's disease, in both of whom unilateral pulsating exophthalmos was present. X-ray examination of the skull revealed defects in the bony wall of the orbit in both cases. These are reported here as further examples of this interesting condition.

Case Reports

Case 1.—This was a young man, aged 26 years, with no previous history of ill-health. He was admitted to the Department of Neurosurgery, Royal Infirmary, Cardiff, on March 27, 1955. About 12 months previously he had noticed that the vision in his left eye was blurred, but this visual defect had not progressed. Six months later, while combing his hair, he became aware that his left eye was pulsating. He was certain that before this the eye had been normal except for the poor visual acuity. There was no complaint of headache, but he had occasional attacks of sharp pain, shooting in character, above the left eye. There was no subjective impression of pulsation of the eye, and there was no history of diplopia.

On examination the left eye was slightly prominent and showed definite backward and forward movement which was synchronous with the pulse. Compression of the left carotid artery lessened the amplitude of the pulsations and also caused some enophthalmos; compression of the right carotid reduced the amplitude of the pulsations only. Compression of the jugular veins increased the degree of exophthalmos. The fundi were normal; there was no engorgement of the retinal veins on the left side and the degree of venous pulsation was normal. Visual acuity on the left was impaired, but was normal on the right. The visual fields were full and external ocular movements were normal. No bruit was heard on auscultation. Neurological examination did not reveal any abnormal physical signs. He had a mild degree of generalized neurofibromatosis, as indicated by the presence of several café-au-lait spots and small violaceous cutaneous neurofibromata scattered over the trunk.

FIG. 1a and b (Case 1).—Postero-anterior and lateral radiographs showing the partial absence of the roof and the posterior wall of the left orbit.

Radiological Examination of Skull.—Postero-anterior projections showed a uniform translucency of the left orbit in sharp contrast to the normal appearances on the right (Fig. 1a). There were no shadows corresponding to the orbital plate of the frontal bone, or the lesser wing and orbital plate of the sphenoid bone. The oblique sphenoid line in the left orbit was absent. There was a large orbital extension of the frontal sinus on the left and the ethmoidal cells medial to the orbit were narrow. The corresponding cells on the right were large and prominent. There was no bulging of the squamous temporal bone. Lateral views (Fig. 1b) showed an abnormally deep pituitary fossa with a steep anterior wall which was lacking in definition. The posterior part of the roof and the posterior wall of the left orbit were absent. The right anterior clinoid process was large and

pneumatized; the left anterior clinoid process was not seen. In the basal view the anterior wall of the left middle fossa was not apparent. The foramina spinosa were identified, but the other basal foramina were poorly defined on both sides. The lateral part of the body of the sphenoid was ill-defined.

The pressure of the lumbar cerebrospinal fluid was 120 mm. Manometrics, chemistry, and the cytology of the fluid were normal. The blood Wassermann and Kahn reactions were negative.

Electroencephalography.—The record showed fairly high voltage alpha rhythm at 10 c./sec. arising in the occipital lobes. Short runs at 7 c./sec. theta waves also occurred in all leads and were augmented by hyperventilation.

FIG. 2a and b (Case 1).—Antero-posterior and lateral angiograms showing the abnormally-shaped and medially-placed carotid siphon.

Left Percutaneous Carotid Angiography.—The carotid siphon (Figs. 2a and b) was abnormal in shape and position, lying more medially than normal. The C2 and C4 portions of the siphon were abnormally long, with the C3 portion lying well forwards. The whole siphon lay more anteriorly and medially than normal while at one point the medial border of the siphon lay 1 cm. to the right of the midline. The C1 portion of the siphon was directed upwards and laterally, the bifurcation being 1 cm. anterior to the posterior clinoids, and on a level with them. Radiological examination of the lumbar spine, the right femur, and the left humerus revealed no abnormality.

Operation.—Through a left frontal craniotomy (C.L.) performed on April 6, 1955, the anterior and middle cranial fossae were explored. The anterior fossa was small from before backwards but otherwise appeared normal. There was no evidence of tumour on the wing of the sphenoid. The left olfactory tract and optic nerve were normal in position and appearance, but the internal carotid artery was not seen. On palpating the anterior wall of the middle fossa there was a soft area about 3 to 4 cm. in diameter where the bone was deficient. This region was also explored extradurally and revealed the orbital fat presenting immediately under the dura mater.

The patient made an uneventful recovery after operation and was subsequently discharged from hospital. Periodic follow-up examinations have shown no increase in the degree of exophthalmos or pulsation.

Case 2.—This was a woman aged 34 years who was admitted to the Department of Neurosurgery on June 28, 1955. Unfortunately, owing to the patient's mental state and the fact that her husband had little command of the English language, it was not possible to obtain an adequate history. The patient, the mother of seven children, had recently left hospital after her latest confinement. About nine months before this, a progressive change in her mental state was noticed. She became apathetic and lost interest in her children of whom she was formerly very fond and of whom she took great care. She lost her personal pride, became slovenly in dress and cleanliness. She complained of frontal headache and at times was confused and disorientated. The deterioration in her mental state became so marked that she was admitted to a local hospital on June 5, 1955.

On examination she was drowsy, apathetic, and it was not possible to engage her in conversation. She had several café-au-lait spots and many cutaneous and subcutaneous neurofibromata scattered over her body. The skull was asymmetrical with a well-marked bulge in the left temporal region. The left eye was slightly prominent and on close examination pulsation of the globe could be detected, which was synchronous with the pulse. Compression of the carotid arteries and the jugular veins produced the same effects as described in the first case. The fundi were normal. A sixth nerve paresis was present on both sides and an upper motor neurone facial weakness on the left. Slight weakness was detected in the left upper limb and the reflexes here were slightly increased. Both plantar responses were extensor, but otherwise no abnormal signs were found in the lower limbs.

FIG. 3a and b (Case 2).—Postero-anterior and lateral radiographs to show the marked elevation of the floor of the anterior fossa and the defect in the posterior wall of the orbit on the left.

A clinical diagnosis of von Recklinghausen's disease with a right frontal tumour and congenital bone defects of the left orbit was made.

Radiological Examination of Skull.—Postero-anterior projections (Fig. 3a) showed that the floor of the anterior fossa on the left was very markedly elevated, particularly laterally. In the tilted P.A. view the line of the floor of the anterior fossa on the left lay above the superior border of the left orbit. The oblique line of the sphenoid in the left orbit was displaced laterally. There was well-marked outward bulging of the left squamous temporal bone. The left orbit was enlarged in all diameters when compared with the right. The normal details of the posterior wall of the left orbit were missing. The anterior clinoid process on the left could be identified and the lesser wing

FIG. 4a and b (Case 2).—Antero-posterior and lateral angiograms to show the abnormal position of the carotid siphon.

of the sphenoid traced from this, laterally and steeply upwards to fuse with the line of the anterior fossa at and above the superior border of the orbit. In the lateral view (Fig. 3b) the anterior wall and floor of the middle fossa were displaced markedly forwards and slightly downwards. The sella turcica was not abnormal.

Electroencephalography.—The record showed a diffuse theta rhythm at 6 to 7 c/sec. throughout. There was very little 8 c/sec. alpha rhythm. There was no apparent focus, but the record indicated diffuse abnormality, probably indicating a deep lesion.

Left Percutaneous Carotid Angiography.—The carotid siphon (Fig. 4a and b) was abnormal in shape and position, lying more medially than normal. The medial border of the siphon reached the midline at one point. The C1 portion was directed upwards and laterally. The bifurcation lay 0.5 cm. above and anterior to the posterior clinoid process.

Subsequent ventriculography confirmed the presence of a right frontal space-occupying lesion.

Operation.—Through a right frontal craniotomy (A.D.) performed on July 6, 1955, an extensive infiltrating glioma was partially removed. Histological examination showed that the tumour was a piloid astrocytoma.

In spite of the fact that an adequate internal decompression was obtained at operation, there was no improvement in the patient's condition. She died in coma on July 20, 1955.

Necropsy.—This confirmed the clinical findings and the extent of the bone defects as shown in radiological examination. The pituitary gland was normal and there was no evidence of an intracranial or intra-orbital neurofibroma.

Discussion

Exophthalmos in cases of von Recklinghausen's disease may be due to an orbital tumour, a glioma of the optic nerve, or be the result of congenital orbital bone defects. A correct diagnosis is obviously of considerable importance. If the exophthalmos is due to congenital absence of part of the orbital wall and the true nature of the lesion is not recognized the patient may be led into an unnecessary surgical procedure. In our first patient a mistaken diagnosis of a vascular sphenoid wing meningioma was made and a transfrontal exploration carried out.

There is no doubt that these congenital defects should be suspected on clinical examination, for, as stressed by Robertson (1949), "the condition is so characteristic that a mistake is unlikely once the condition is known". The final recognition of the disorder, however, rests on an adequate radiological examination of the skull. The appearances are distinctive and are not likely to be confused with any other condition. Contrast studies are not essential, although they may give valuable additional information. While the extent of the bony abnormality may vary from case to case, characteristically there is an absence of the bony detail in the posterior wall of the orbit due to a partial absence of the orbital plate of the frontal bone, and a similar failure in development of the greater and lesser wings of the sphenoid bone. The body of the sphenoid also is usually involved, as shown by the abnormal shape of the sella turcica. In addition the squamous temporal bone often bulges. The affected orbit is larger than normal, the ethmoidal air cells on the same side are usually narrow and the maxillary antrum small.

A point of importance in the differential diagnosis of these bone defects is their clear-cut nature, which contrasts with the ill-defined margins of bone defects produced by tumour erosion.

Contrast studies have not been reported on many of these patients. Robertson (1949) reported a case in which an orbital encephalocele was demonstrated by air encephalography. Petit-Dutailis *et al.* (1949) and Elslo and van der Zwan (1952) have reported cases and described the findings on angiography. In both of our patients angiography was carried out and an abnormality revealed. The most striking feature was the medial displacement of the whole of the carotid siphon, which also lay more anteriorly than normal. These features were most obvious in our first case, where in addition the C2 and C4 portions of the siphon were elongated. The size of the internal carotid artery was not abnormal in either of our cases.

In the case described by Elslo and van der Zwan (1952) the carotid siphon was also abnormal in shape and position with elongation of the C2 and C4 portions. Unfortunately the relationship of the siphon to the midline was not mentioned. These appearances were interpreted as an aneurysm. The authors felt that the protrusion and pulsation of the eyeball were due to this, and that the enlargement of the sella and the bone defect of the orbit might be due to the continuous pulsation of the aneurysm and contents of the orbit. We do not think that the abnormal carotid siphon demonstrated in our cases can be classified as an aneurysm. In our opinion the bony anomaly is the primary lesion and the unusual vascular pattern a secondary manifestation of this defect. The dislocation of the carotid siphon to the midline is the result of a failure in development of the corresponding side of the body of the sphenoid, and the forward displacement, and elongation of parts of the siphon, to the absence of the posterior wall of the orbit.

It seems probable that some of these cases never develop symptoms which call for treatment and go through life without the condition being recognized. Certainly the degree of pulsation of the globe in our two cases was such that there was no subjective disturbance and the exophthalmos was slight. In Case 2 the bony anomaly was only an incidental

finding in the presence of another more serious lesion. In those patients with a marked degree of exophthalmos transfrontal repair of the orbit roof, as described by Dandy (1929), would appear to be the operation of choice. However, marked or progressive exophthalmos, occurring in a patient with von Recklinghausen's disease, should call for full investigation even in the presence of undoubted congenital bone defects of the orbital wall. Intracranial tumours are not uncommonly associated with neurofibromatosis, as in our second case, and the orbital wall defect may then act as a natural decompression in the presence of a rising intracranial pressure. It is advisable, therefore, if a surgical repair of the orbital wall is contemplated, to carry out preliminary angiography or air studies.

Summary

Two cases of von Recklinghausen's disease with pulsating exophthalmos due to congenital bone defects in the orbital wall are reported.

The radiological appearances are described and the abnormality revealed by cerebral angiography is discussed.

Treatment is briefly discussed and the importance stressed of carrying out contrast studies before surgical treatment in patients with progressive exophthalmos.

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