Visual Loss Secondary to a Giant Aneurysm in a Patient with Tuberous Sclerosis

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ABSTRACT: We report a fifty three year old man who presented with progressive visual loss. Investigation disclosed a giant aneurysm with optic nerve compression, which was successfully treated surgically. The patient had typical skin lesions of tuberous sclerosis, but did not suffer from a seizure disorder or mental retardation. In the past he had had an enucleation for glaucoma secondary to a retinal phakoma.

Cerebral aneurysms are discussed in relation to the tuberous sclerosis disease process. After review of the literature, we propose that the aneurysms are secondary to dysplastic change in the vessel walls, this is in keeping with the abnormal tissue proliferations in this disease.

RÉSUMÉ: Nous rapportons le cas d'un homme de 53 ans, atteint d'un déficit visuel progressif, chez qui l'investigation révèle la présence d'un anévrysme géant comprimant le nerf optique, qui fut opéré avec succès. Porteur de lésions cutanées typiques de sclérose tubéreuse, il n'avait ni déficience mentale, ni passé comitial. Par contre, il avait subi une énucléation pour glaucome secondaire à un phacome rétinien.

La relation des anévrysmes cérébraux et de la sclérose tubéreuse est ensuite discutée. Après revue de la littérature, nous proposons que ces anévrysmes cérébraux sont le résultat de modifications dysplasiques au niveau de la paroi des vaisseux. Cette hypothèse cadrerait avec la prolifération tissulaire anormale qui caractérise cette affection.

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Tuberous sclerosis was described in 1880 by Bourneville as potato like thickenings of the cerebral cortex, but von Recklinghausen was, in fact, the first to describe the syndrome in 1862. He published a case of a neonate with multiple sclerosis of the brain and myomata of the heart. Since then, tuberous sclerosis has become known as an hereditary disorder affecting a multiplicity of body systems. It presents as a classical clinical triad consisting of mental retardation, seizure disorders and skin lesions.

The purpose of this report is to discuss the possible association of aneurysms in the cerebral circulation in patients with tuberous sclerosis. This is based on a case of visual failure in a middle aged man.

Case Report

A fifty three year old man was admitted for evaluation of decreased vision of the left eye. He had first come to medical attention twelve years earlier. At that time he had a four month history of progressive central visual loss with acuity of 20/200 and an exophytic retinal phakoma. This was later confirmed by pathological examination, following enucleation for glaucoma secondary to the tumour. A diagnosis of tuberous sclerosis was made on clinical grounds based on numerous skin lesions and the retinal phakoma.

Four months prior to this admission, the patient noted a painless, gradually progressive blurring of vision in his remaining eye, which was exacerbated by bright light. Dyschromatopsia later developed. He was otherwise well.

His past history was unremarkable, with no seizures and with normal mental function. Family history was also negative for mental retardation, orbital tumours or tuberous sclerosis. His father had petit mal seizures as a child.

Physical examination revealed a healthy male in no distress. A dermatological examination showed periungual and subungual fibromas, shagreen patches on the buttocks and back (pathologically confirmed), numerous collagen nevi, facial angiofibromata, (primarily nasal and malar), and hypopigmented "ash leaf" macules that were demonstrated on the abdomen with a Wood's lamp. There was a right orbital prosthesis. The visual acuity in the left eye was 20/60 corrected. There was central dyschromatopsia. The remainder of the ophthamologic, neurologic and general examination was normal.

Skull x-rays disclosed a mottled sclerotic vault with scattered intracranial calcifications. An abdominal ultrasound showed a single renal stone, but no evidence of abdominal parenchymal cysts. IVP was normal, aside from the abovementioned stone. ECG and 2-D echocardiogram were normal as well.

A contrast enhanced CT of the head revealed subependymal calcifications in the lateral ventricles and scattered hemispheric calcifications.

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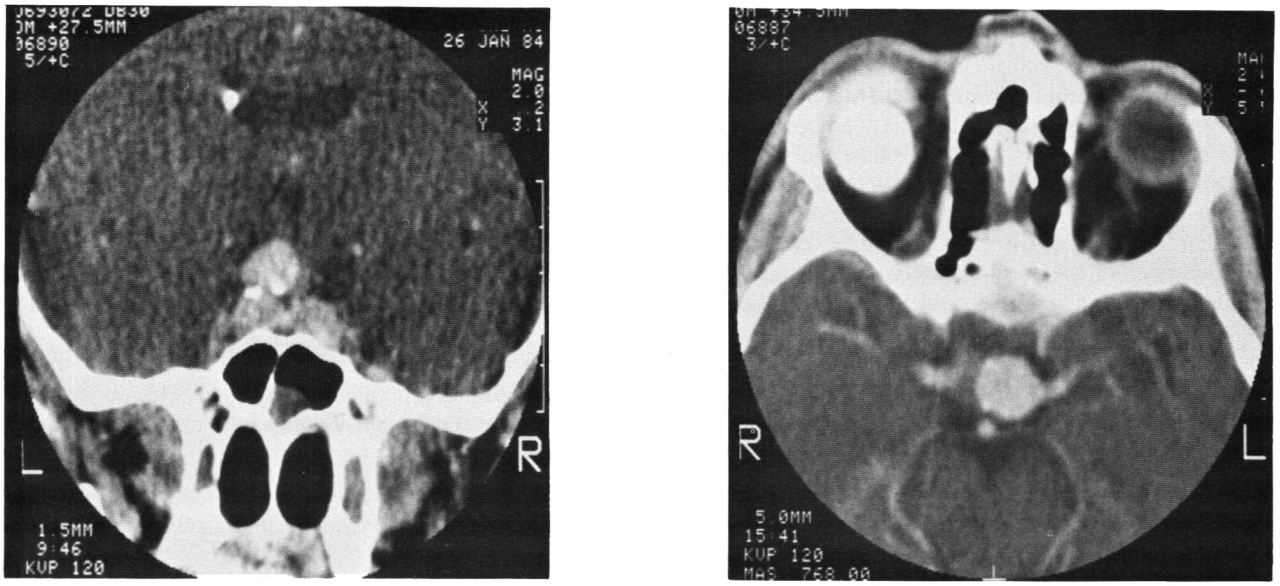

Figure 1 — a) Coronal CT of the head with enhancement showing the suprasellar aneurysm and subependymal calcification b) Axial CT.

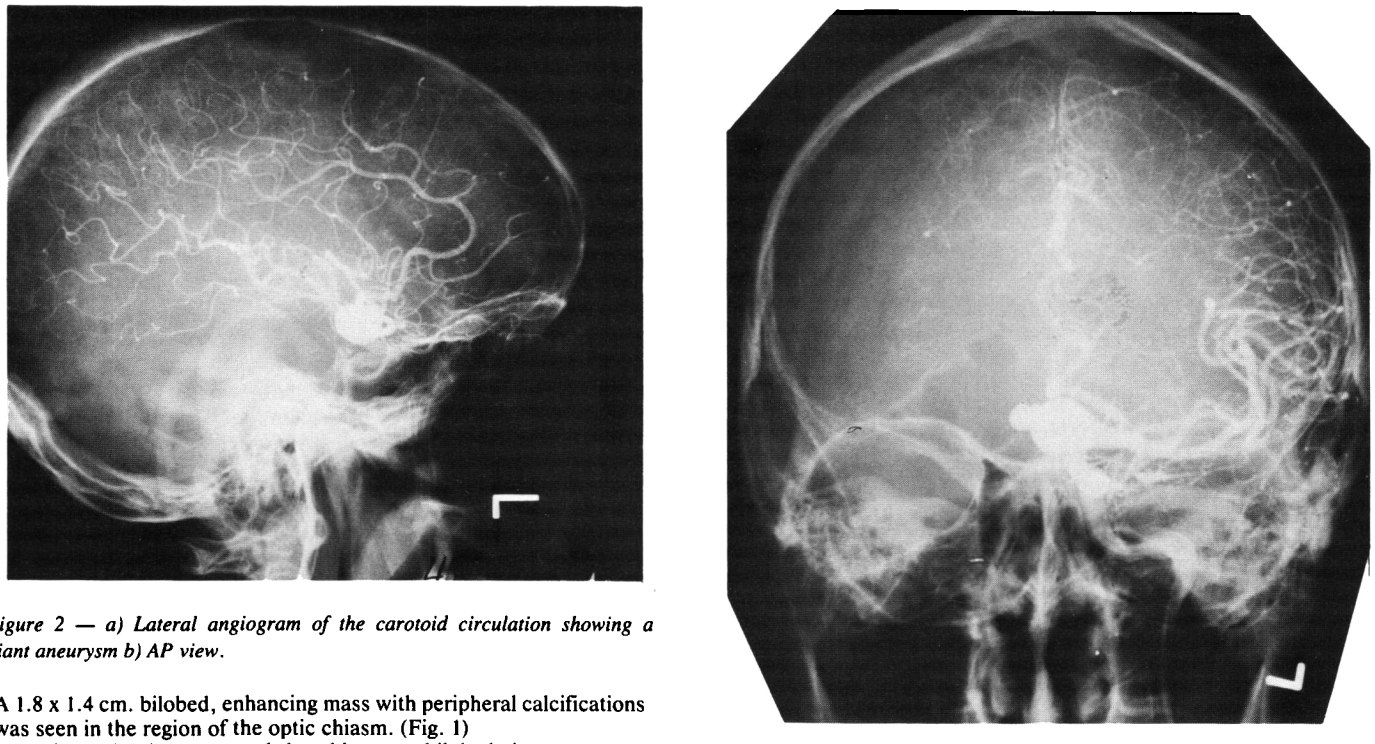

Figure 2 — a) Lateral angiogram of the carotoid circulation showing a giant aneurysm b) AP view.

A 1.8 x 1.4 cm. bilobed, enhancing mass with peripheral calcifications was seen in the region of the optic chiasm. (Fig. 1)

Angiography demonstrated that this was a bilobed giant aneurysm stemming from the supraclinoid portion of the left internal carotid artery. The bilateral carotid and vertebral angiograms were otherwise normal. (Fig. 2)

At surgery, an aneurysm was found arising from the distal segment of the left internal carotid artery. It passed under and compressed the left optic nerve. The aneurysm was clipped and decompressed.

The post operative course was unremarkable. At the time of follow-up the visual acuity had improved to 20/40.

Discussion

Tuberous sclerosis is considered a disease of abnormal proliferations of tissue, indigenous to the involved organ. In many tissues the related cells are similar to the embryonic undifferentiated cells. As the affected individual matures, these cells may differentiate in unusual directions resulting in the many different lesions inherent to this disease. (Bender and Jinis, 1982). The giant aneurysm in this case may be a manifestation of abnormal vasculature found in tuberous sclerosis.

Pathologically, this disease displays many areas of abnormal vasculature in the body. Angiomyolipomas of the kidney have vessels that are large, thick walled and are morphologically similar to arteries. They do not have an arterial elastic tissue pattern and they frequently have eccentric lumena (Robbins et

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al., 1979; Bender and Jinis, 1982). In addition, the spleen is the site of the angiomatous dysplasia and malformation (Gomez, 1979; Bender and Jinis, 1982). Gomez (1979) also describes abnormal vasculature in the hepatic lesions of tuberous sclerosis.

Major vessels are rarely involved in this condition. Larbie et al. (1971) describe an aneurysm of the descending thoracic aorta in a two and half year old boy, with skin lesions of tuberous sclerosis. An autopsy revealed cerebral tuberous sclerosis, cardiac rhabdomyoma, a polycystic kidney and a saccular aneurysm of the aorta. There were two more aortic aneurysms in formation. A microscopic examination disclosed an almost complete disappearance of all normal structures. The aortic wall contained fragmented dystrophic elastic fibres in the intima. The media had lacunes containing fragmented elastic fibres and edema. At the level of the aneurysms in formation there was considerable elastic dystrophy interupting the media but permitting distension of the vessel wall. There are three other reported cases of aortic aneurysms in tuberous sclerosis (Gomez, 1979).

Abnormalities of the cerebral blood vessels have also been shown radiologically. Hilal et al. (1971) and Medley et al. (1976), discuss the occurence of cerebral vascular occlusive disease in tuberous sclerosis. In patients with tuberous sclerosis and giant cell astrocytoma, angiography shows a characteristic abnormality. In the late arterial phase there is a berry like aneurysmal dilatation and an absence of early draining veins. This may help to differentiate from a more malignant tumour (Herz et al., 1978).

There are three case reports in the literature describing cerebral aneurysms in tuberous sclerosis. Snowdon (1974) described a twenty four year old man who died after a subarachnoid hemorrhage. At necropsy, the patient was found to have had a middle cerebral artery saccular aneurysm, tuberous sclerosis of the brain and polycystic kidneys. The author, however, does not discuss the association of polycystic kidneys and cerebral aneurysms. Unfortunately, there is also no discussion of the histology of the aneurysm wall.

The second case of tuberous sclerosis (Davidson, 1974) is of a child with bilateral fusiform aneurysms of the intracranial carotid arteries. The patient presented with a unilateral visual disturbance. She was shown to have had bilateral involvement in the region of the carotid siphon. On the affected side she also had involvement of the origin of the ophthalmic artery. The patient was not treated surgically and at the time of the report had been stable for eight years.

The third report is of a seventeen year old girl who presented with reduced visual acuity, an altitudinal defect and a relative afferent pupillary defect in the left eye. The patient had a previous diagnosis of tuberous sclerosis. She had a giant aneurysm in the left supraclinoid and cavernous portion of her internal carotid artery, which was treated surgically (Beall et al., 1983).

We propose that the etiology of the intracranial aneurysms in tuberous sclerosis is dysplasia of the vascular wall. This would correspond with the general observation that the disease is one of abnormal cell proliferation and hamartomatous change in many tissues of the body. Unfortunately, none of the four cases now in the literature have pathological studies to verify this.

A case can be made to suggest that the dysplasia seen in the patient with aneurysms of the aorta is similar to those involving the cerebral circulation. Other evidence points to histological studies of abnormal vessels in the lesions of renal angiomyolipomas, liver, spleen and giant cell astrocytomas, which also have similar dysplasia with hamartomatous change. In addition, there is evidence from angiographic studies that the lesions are similar.

Aneurysms of the intracranial and extracranial vessels may be more common than is now realized. This abnormality perhaps should now be considered another feature of the tuberous sclerosis complex.

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References

Beall S, Delaney, P (1983) Tuberous sclerosis with intracranial aneurysms. Arch Neurol 40: 826-827.

Bender BL, Jinis EJ (1982) The pathology of tuberous sclerosis. Path Annual Part 1, (vol 17), 339-383.

Bourneville DM (1880) Contribution a l'étude de l'idiotie sclerose tubereuse des circonvolutions cerebrales; idiotie et épilepsie hemiplégique. Arch Neurol (Paris) 1: 81-91.

Davidson S (1974) Tuberous sclerosis with fusiform aneurysms of both internal carotid arteries maintained by unilateral visual loss and papilledema. Bull Los Ang Neur Soc 39 (3): 128-132.

Gomez MR (1979) Other visceral, vascular and osseous lesions. Tuberous Sclerosis. ed. M Gomez, New York: Raven Press, pp 171-192.

Hilal SK, Solomon GE, Gold, AP, Carter S (1971) Primary cerebral arterial occlusive disease in children. Radiology 99: 87-93.

Herz PA, Liebskind A, Rosenthal A, Schechter MM (1975) Cerebral angiographic changes associated with tuberous sclerosis. Radiology 115: 647-649.

Larbie F, Loire R, Guibaud B, Weill B (1971) Observation clinique et anatomique aux anévrysm de l'aorte au cours de la sclereuse tubereuse de Bourneville. Arch Fr Pediat 28: 975-984.

Medley BE, McLeod RA, Houser OW (1976) Tuberous sclerosis. Seminars in Roentgenology. vol xi (1).

Prompitak A, Maberley AL, Shea M (1973) An abortive case of tuberous sclerosis without mental deficiency or epilepsy in an adult. Amer Jour of Ophthamology 76 (2): 255-259.

von Recklinghausen F (1862) Ein Herz von einem Neugeborenen welches mehrere Theils nach aussen, Theils nach den Hohlen prominirende Tumoren (Myomen) trug. Verh Ges Geburtsch Monatschr Beburtsk 20: 1-2.

Robbins TD, Bernstein J (1979) Tuberous Sclerosis. ed. M Gomez New York: Raven Press pp 171-192.

Snowdon JA (1974) Cerebral aneurysm, renal cysts and hanartomas in a case of tuberous sclerosis. Br Jr of Urology 46: 583.