

Case Report

Tuberous sclerosis with giant internal carotid artery aneurysm resulting left third nerve palsy: A case report

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Abstract

Tuberous sclerosis is an autosomal dominant hereditary condition with many varied forms of clinical presentation. The most frequent cutaneous findings in tuberous sclerosis include multiple angiofibromas, hypopigmented macules, periungual fibromas and shagreen patch. It is characterized by the development of hamartomatous growths in many organs. We present a case of tuberous sclerosis with a giant left internal carotid artery aneurysm causing pulsatile proptosis, left sided ptosis, anisocoria and papillary mydriasis indicative of left third cranial nerve palsy.

Key words: Tuberous sclerosis, internal carotid artery aneurysm, third nerve palsy

Introduction

Tuberous sclerosis is a neurocutaneous syndrome that is inherited in an autosomal dominant pattern or can arise from a spontaneous mutation. Mutations in the TSC1 and TSC2 genes are responsible for tuberous sclerosis.¹ With advances in molecular genetics, the TSC2/PKD1 contiguous gene syndrome has been described. TSC2 and PKD1 genes share a common location at chromosome 16p13.3. Manifestations of tuberous sclerosis involve multiple organ systems and it is classically characterized by the triad: mental retardation, seizures, and facial angiofibromas.² Other well-known features include cortical tubers, subependymal giant cell astrocytoma, cardiac rhabdomyomas and hypopigmented macules.² Cerebral aneurysms are less commonly described and usually manifest with cranial nerve deficits.³ Once diagnosed, aneurysm occlusion with internal carotid artery clip ligation or endovascular embolization is the treatment of choice.

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Case report

A 15-year-old male had been diagnosed with tuberous sclerosis with a giant left internal carotid artery aneurysm at 13 years of age in 2011. He presented with the features of raised intracranial pressure with left sided visual impairment for 4 months in April 2011. There was no seizure. Family history was negative. Examination showed hypomelanotic macules and shagreen patches on limbs, face and trunks; facial angiofibromas, left sided ptosis with pulsatile proptosis and left sided dilated pupils (Figure-1, Figure-2).



Figure-1: Multiple adenoma sebaceum in face



Figure-2: Showing multiple ash-leaf spots in face and trunk.

Visual acuity was 6/6 on the right eye and there was no perception of light on left eye. Fundoscopy revealed left sided optic atrophy. There was no motor deficit. Computed tomography scan of brain demonstrated a large (6cm x 4.2cm) hyperdense extra-axial rounded mass in the left middle cranial fossa. There were calcified cortical tubers and subependymal nodules. Magnetic resonance imaging and Magnetic resonance angiography of brain were done and found a huge fusiform aneurysm in the terminal part of left internal carotid artery.

Four vessel digital subtraction angiography of cerebral vessels was performed which revealed a hugely dilated fusiform aneurysm extending from cavernous part to bifurcation of left internal carotid artery. After occlusion of left common carotid artery, right internal carotid artery angiogram revealed a patent anterior communicating artery through which the left sided middle carotid artery circulation is well maintained. Abdominal ultrasonogram revealed no abnormality. After this, surgery was planned and he underwent ligation of left internal carotid artery with simultaneous intracranial trapping of the aneurysm on May 2011. Post-operatively his ptosis and pulsatile proptosis improved. Vision in the left eye has not been recovered. Follow-up computed tomography scan of brain was done on September 2013 which showed a thrombosed left internal carotid artery aneurysm (Figure-3).

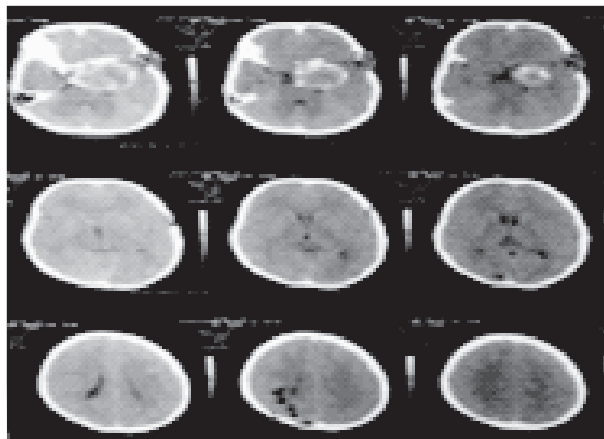


Figure-3: Post operative follow-up Computed tomography scan of brain showing a thrombosed left internal carotid artery aneurysm

Discussion

Tuberous sclerosis was first described by Von Recklinghausen in 1862⁴ and then named by the French pediatric neurologist Désiré-Magloire Bourneville in 1880⁵. Its name is derived from the Latin word “tuber” for “swelling” and the Greek word “skleros” for “hard”. It has long been classified as a neurocutaneous phakomatosis, inherited in an autosomal dominant pattern, with an incidence of 1 per 30,000 in the general population and a birth incidence of 1 per 5800.⁴

Manifestations of tuberous sclerosis vary widely, involve multiple organ systems, arise at distinct stages of development, and have highly variable phenotypic penetrance. Criteria for making the diagnosis of tuberous sclerosis have been categorized into major and minor features, of which two major, or one major and two minor features are diagnostic (Table-I)²

Table -I: Criteria for tuberous sclerosis²

Major Criteria	Minor Criteria
Facial angioma	Multiple pits in dental enamel
Ungual fibroma	Hamartomatous rectal polyps
Shagreen patch	Bone cysts
Hypomelanotic macule	Multiple renal cysts
Internal carotid artery tuber	Gingival fibromas
Subependymal nodule	Aneurysms
Subependymal giant cell astrocytoma	
Retinal hamartoma	
Cardiac rhabdomyoma	

Patients typically present with seizures or dermatologic findings. More common presentations of tuberous sclerosis include facial angiofibromas (adenoma sebaceum), hypopigmented macules (ash-leaf spots), renal angiomyolipomas, cardiac rhabdomyomas, pulmonary lymphangiomyomatosis, cortical tubers. Less common presentations include aneurysms, dental enamel pits, renal cysts and hamartomatous rectal polyps.²

While seizures and cerebral lesions are very common in tuberous sclerosis, arterial aneurysms are infrequently encountered. Smaller caliber vessels appear to be more prone to involvement, but patients have presented with medium and large vessel involvement as well. Locations include the thoracic and abdominal aorta,⁶ pulmonary arteries,⁷ iliofemoral arteries,⁸ carotid arteries,⁹ and cerebral arteries.³ Vascular histopathology has demonstrated abnormal architecture, including fragmented or deficient elastic fibers, mucopolysaccharide deposition, dense fibrous tissue, and calcified internal carotid artery.^{10,11}

Only 17 prior cases of intracranial aneurysms in tuberous sclerosis have been reported in the literature.³ Age at presentation varies widely, from 5 months to 53 years, and neither gender is predominant. Examination findings of tuberous sclerosis patients with intracranial aneurysms are many. As in our patient, cranial neuropathies may be the presenting symptom. Other signs and symptoms of cerebral aneurysms include visual loss secondary to optic nerve compression,^{12,13} cavernous sinus syndrome from third and sixth cranial nerve compression,¹⁴ direct compression on cavernous sinus causing pulsatile proptosis, and more diffuse deficits such as hemiparesis secondary to a mid-basilar artery aneurysm.¹¹ Intraventricular hemorrhage from aneurysm rupture has also been reported in the literature.¹⁵ Most commonly, however, vascular lesions are incidental findings on routine imaging obtained to monitor parenchymal lesions.

Due to the paucity of reported cases, treatment of cerebral aneurysms in tuberous sclerosis patients has not been well described. One case of an extracranial aneurysm of the right carotid artery bifurcation was treated with open external carotid artery ligation and reanastomosis of the distal common carotid to the internal carotid arteries.¹⁰ As balloon occlusion test revealed adequate circulation from right internal carotid artery in our case, anastomosis was not required.

Due to lack of facility of endovascular procedures in our country we arranged for ligation proximal and distal to the aneurysm sac to prevent anterograde and retrograde filling. Although 3rd nerve palsy improved, visual problem did not improve.

Intracranial aneurysms are a rare finding in tuberous sclerosis, but should be considered in patients with new cranial neuropathies. Neither surgical nor endovascular outcomes have been well described in the literature. We report the successful surgical occlusion of a large fusiform aneurysm extending from the left cavernous to distal internal carotid artery in a 13 year old male with tuberous sclerosis. Result is early resolution of the third cranial neuropathy.

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