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Case Report

Coil embolization of an intracranial aneurysm in an infant with tuberous sclerosis complex: A case report and literature review

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Abstract

Background: Tuberous sclerosis (TS) is a multiorgan neurocutaneous syndrome. Vascular manifestations are often extracranial. There is a paucity of cases involving TS combined with intracranial aneurysms reported in the literature. As a result, treatment has not been well described.

Case Description: We report the case of a 13-month-old female infant with a prior diagnosis of TS that was found to have new onset of left eye ptosis, anisocoria, and papillary mydriasis indicative of left third cranial nerve palsy. A magnetic resonance angiogram (MRA) of the brain revealed a left internal carotid artery (ICA) aneurysm. Endovascular embolization was determined to be the best option for treatment. After a successful balloon test occlusion with neuromonitoring, the left internal carotid artery was sacrificed via coil embolization.

Conclusions: This is only the third case report of endovascular coil embolization of an intracranial aneurysm in an infant with TS. We report no complications during the procedure, and the patient was discharged with resolving left third cranial nerve palsy. Neither surgical nor endovascular outcomes have been well described in the literature. Follow-up on this patient will be useful for establishing protocols of treatment.

Key Words: Hamartin, intracranial aneurysm, tuberin, tuberous sclerosis

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INTRODUCTION

Tuberous sclerosis (TS) 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 TS.[19] Manifestations of TS involve multiple organ systems, and it is classically characterized by the triad: mental retardation, seizures, and facial angiofibromas.[10] Other well-known features include cortical tubers, subependymal giant cell astrocytoma (SEGA), cardiac rhabdomyomas, and hypopigmented macules.[10] Cerebral aneurysms are less commonly described and usually manifest with cranial nerve deficits.[10,13,11,22] Once diagnosed, aneurysm occlusion with surgical clip ligation or endovascular embolization is the treatment goal.

CASE REPORT

Case summary

We present the case of a 13-month-old female infant with a diagnosis of TS who was born term at 37 weeks

gestation to a healthy mother who had an uncomplicated pregnancy. At 3 months of age, she presented with new-onset seizures characterized by leftward head deviation and rhythmic facial, oral, and extremity movements. Further evaluation revealed ash-leaf hypopigmented macules, subependymal tubers on brain magnetic resonance imaging (MRI), and rhabdomyomas on echocardiography. A diagnosis of TS was made and the seizures were treated with levetiracetam for almost 1 year. Subsequently, the patient developed myoclonic activity with an EEG pattern consistent with modified hypsarrhythmic activity leading to the diagnosis of infantile spasms. She was then treated with adrenocorticotropic hormone (ACTH).

The patient presented now at 13 months with new onset of left third cranial nerve palsy, ptosis, and mydriasis, consistent with ipsilateral third cranial nerve palsy. MRI now showed the interval development of a left cavernous sinus lesion suspicious for an aneurysm [Figure 1]. Magnetic Resonance Angiogram (MRA) of the brain revealed an 18-mm-diameter left cavernous internal carotid artery (ICA) aneurysm that further extended into the petrous and distal cervical ICA [Figure 2].

Decision making

Due to the size of the aneurysm and its rapid development, it was decided that the aneurysm required treatment to prevent hemorrhage. Surgical intervention was deemed high risk for an aneurysm in this location at the skull base and within the cavernous sinus. Endovascular treatment was deemed to be the safer approach. However, because of the patient's age, combined stent–coil embolization with subsequent antiplatelet therapy in a child with a seizure disorder was deemed unsafe. Therefore, the treatment plan was to perform a balloon test occlusion (BTO) of the ICA, with or without extracranial-to-intracranial arterial bypass, followed by endovascular occlusion. If she passed the BTO with hypotensive challenge, then immediate coil embolization would be performed to occlude the entire aneurysm sac from the paraclinoid ICA segment distally to the cervical ICA proximally. If she failed the BTO, then a direct common carotid-to-M1 segment direct arterial bypass with radial artery grafting would be performed, followed by endovascular occlusion of the paraclinoid ICA segment distally to the cervical ICA proximally, to treat this aneurysm.

Endovascular technique

Under general anesthesia and using intraoperative neuromonitoring (somatosensory and motor-evoked potentials), the BTO was performed using a 3 × 7 mm HyperForm Occlusion Balloon System (ev3 Neurovascular, Irvine, CA, USA) positioned and inflated in the proximal ICA with 10 min of normotension followed by 10 min of hypotension (systolic blood pressures reduced to 70% of baseline) without changes in neuromonitoring signals [Figure 3]. At this point, it was determined that a surgical common carotid artery-to-M1 bypass would not be needed, and the left ICA was sacrificed by coil embolization positioning thick oversized coils (Penumbra Coil 400; Penumbra, Inc., Alameda, CA, USA) just distal to the aneurysm sac to ensure occlusion of the aneurysm outflow zone to prevent retrograde aneurysm filling from the supraclinoid ICA. Additional Penumbra coils were inserted from distal to proximal, until the aneurysm sac and the cavernous, petrous, and distal cervical segments of the ICA were completely occluded with coils. Serial angiography through the left ICA confirmed complete occlusion and isolation of the aneurysm (Aneurysm Embolization Grade "A"[12] and Raymond-Roy Occlusion "1") from anterograde flow [Figure 4]. The right common carotid artery was then accessed, and angiography confirmed the absence of retrograde aneurysm filling via the anterior

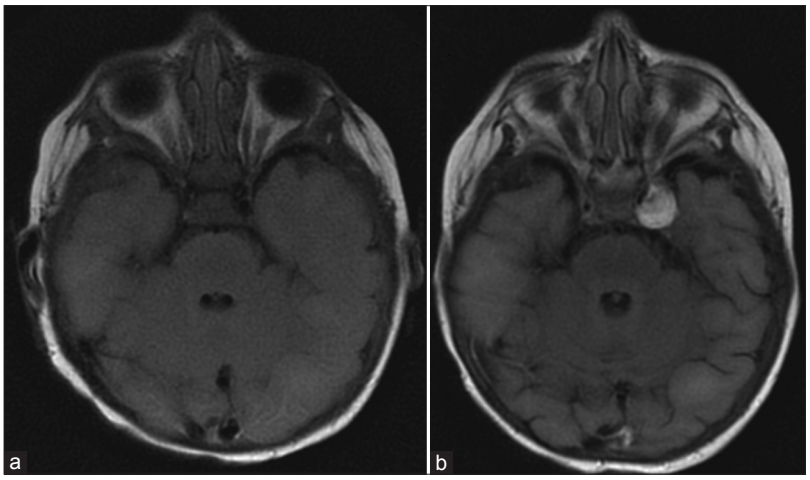

Figure 1: MRI at initial diagnosis (a) Axial T2-weighted fluid-attenuated inversion recovery (FLAIR) MRI showing features of tuberous sclerosis, including white matter radial migration lines and cortical tubers. MRI at presentation of cranial nerve deficits (b) Axial T2-weighted FLAIR MRI showing an 18-mm left cavernous sinus internal carotid artery aneurysm

communicating artery with preserved flow into the left anterior and middle cerebral arteries.

There were no complications during the procedure and no changes in neuromonitoring signals during the embolization. The patient awoke with her baseline neurological examination and recovered in the pediatric ICU. A few days after the procedure, the patient was discharged home in stable condition with resolving third cranial nerve palsy. At 3-month follow-up visit, she was neurologically intact with no third nerve palsy, and MRA demonstrating durable aneurysm occlusion.

DISCUSSION

Tuberous sclerosis was first described by von Recklinghausen in 1862[25] and then named by the French pediatric neurologist Désiré-Magloire Bourneville in 1880.[4] 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.[20]

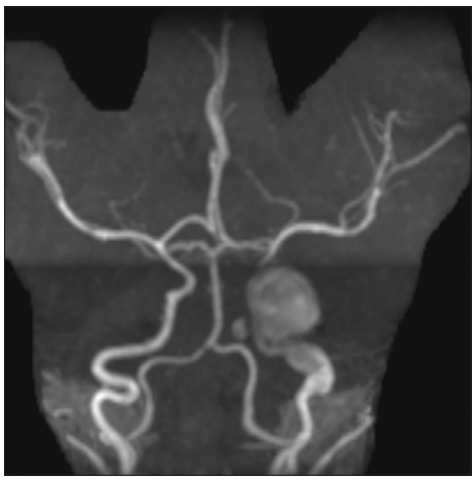

Figure 2: MRA showing the same 18-mm left cavernous sinus internal carotid artery (ICA) aneurysm with extension to the petrous and distal cervical ICA

Clinical manifestations of TS vary widely, involve multiple organ systems, arise at distinct stages of development, and have highly variable phenotypic penetrance. Criteria for making the diagnosis of TS have been categorized into major and minor features, of which two major, or one major and two minor features are diagnostic [Table 1].[10] Patients typically present with seizures or dermatologic findings.[10] More common presentations of TS include facial angiofibromas (adenoma sebaceum), hypopigmented macules (ash-leaf spots), renal angiomyolipomas, cardiac rhabdomyomas, pulmonary lymphangiomyomatosis, cortical tubers, and SEGA.[10] Less common presentations include aneurysms, dental enamel pits, renal cysts, and hamartomatous rectal polyps.[10]

While seizures and cerebral lesions are very common in TS, 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,[18] pulmonary arteries,[7] iliofemoral arteries,[6] carotid arteries,[14] and cerebral arteries.[8,2,24,3,9,5,13,1,14,23,11,6,16,17,15,22,21] Vascular histopathology has demonstrated abnormal architecture, including fragmented or deficient elastic fibers, mucopolysaccharide deposition, dense fibrous tissue, and calcifications.[16,17]

Only 17 prior cases of intracranial aneurysms in TS have been reported in the literature.[8,2,24,3,9,5,13,1,14,23,11,6,16,17,15,22,21] Age at presentation varies widely, from 5 months to 53 years, and neither gender is predominant. Exam findings of TS patients with intracranial aneurysms are many. As in our patient, cranial neuropathies may be the presenting symptom. Other signs and symptoms

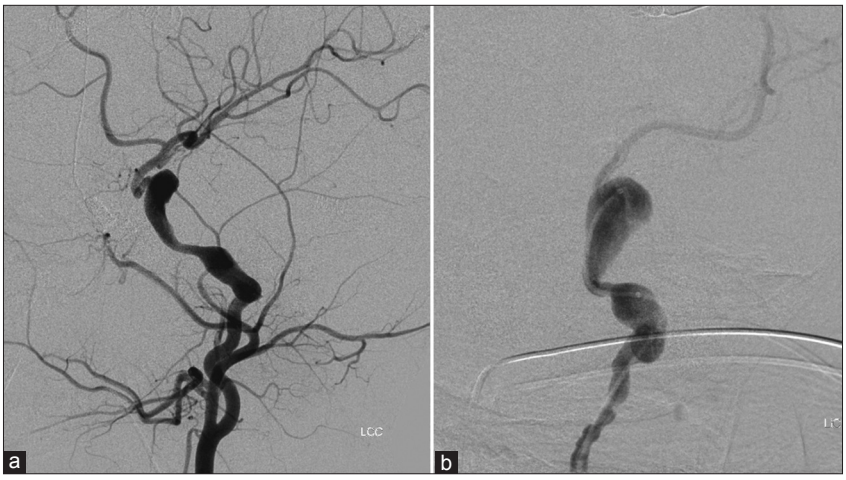

Figure 3: Digital subtraction angiography further characterizing the left internal carotid artery aneurysm (a) Digital subtraction angiography just prior to intervention (b)

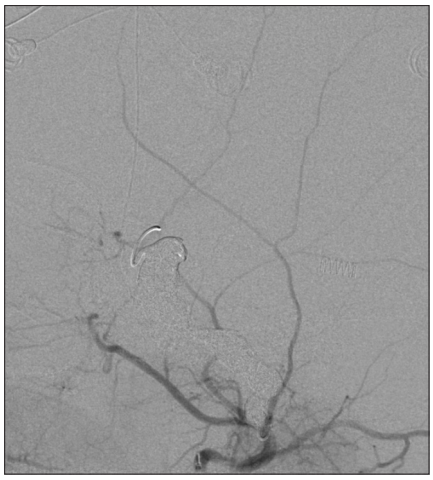

Figure 4: Digital subtraction angiography following coil embolization and complete occlusion of the left cavernous, petrous, and distal cervical internal carotid artery. The contrast seen demonstrates reflux in the external carotid artery branches

Table 1: Criteria for tuberous sclerosis[10]

Major	Minor
Facial angiofibroma	Multiple pits in dental enamel
Ungual fibroma	Hamartomatous rectal polyps
Shagreen patch	Bone cysts
Hypomelanotic macule	Cerebral white-matter radial migration lines
Cortical tuber	Gingival fibromas
Subependymal nodule	Retinal achromic patch
Subependymal giant-cell tumor	"Confetti" skin lesions (groups of small, lightly pigmented spots)
Retinal hamartoma	Multiple renal cysts
Cardiac rhabdomyoma	
Renal angiomyolipoma	
Lymphangiomyomatosis	

of cerebral aneurysms include visual loss secondary to optic nerve compression,[13,11] cavernous sinus syndrome from third and sixth cranial nerve compression,[22] and more diffuse deficits such as hemiparesis secondary to a mid-basilar artery aneurysm.[17] Intraventricular hemorrhage from aneurysm rupture has also been reported in the literature.[24] 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 TS 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 reanastamosis of the distal common carotid to the internal carotid arteries.[16] A similar anastomotic approach was considered for our patient in the form of an extracranial–intracranial direct bypass if she had failed the BTO, suggesting that she would have had inadequate cerebral perfusion with occlusion of the ipsilateral ICA. The patient passed the BTO, however, allowing for coil embolization of the left ICA proximal and distal to the aneurysm sac to prevent anterograde and retrograde filling. Only two prior cases utilizing Gulglielmi detachable coils (GDCs) for total occlusion of intracranial aneurysms associated with TS have been reported.[17,22] Outcomes were satisfactory in the first case, with minor complications including a groin hematoma requiring transfusion, headaches likely from dural irritation and inflammation, transient fifth cranial nerve palsy, and microembolic infarctions of the cerebellum and occipital poles; ultimately, this patient had no permanent neurologic deficits at 12-month follow-up.[17] In the second case, outcomes were good with resolution of right-sided ptosis and sixth cranial nerve palsy, but residual pupillary dilation and poor reactivity.[22]

CONCLUSIONS

Intracranial aneurysms are a rare finding in TS, 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 endovascular coil occlusion of a large fusiform aneurysm extending from the left cavernous to distal cervical ICA in a 13-month-old female infant with TS. Early resolution of the third cranial neuropathy without aneurysm recurrence suggests that this technique can result in excellent treatment outcomes.

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