Case Report

Successful endovascular treatment of spontaneous intrarenal pseudoaneurysm in a case of tuberous sclerosis ✤

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A B S T R A C T

Tuberous sclerosis is a complex disorder which has multisystem involvement and varied clinical manifestations. Almost half of the patients have associated angiomyolipoma which contains fat, vascular and smooth muscle components. Spontaneous pseudoaneurysm formation is a complication of angiomyolipoma. Here we present a case of a female child presenting with hematuria who after thorough clinical and radiological investigations was diagnosed as a case of tuberous sclerosis with right intrarenal pseudoaneurysm. She was successfully treated with endovascular coil embolization.

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Introduction

Tuberous sclerosis also known as tuberous sclerosis complex or Bourneville disease is a rare autosomal dominant neurocutaneous syndrome associated with TSC1 and TSC2 mutations. The absence of these tumor suppressor genes, either hamartin (TSC1 gene) or tuberin (TSC 2 gene) results in loss of inhibition of mammalian target of rapamycin (mTOR) pathway and leads to multisystemic hamartomas [1,2].

It can affect any organ but mostly involves the brain, skin, eyes, heart, kidneys, and lungs and has variable clinical presentations based on the organs involved. In infants, cortical tubers and cardiac rhabdomyomas are commonly seen, whereas in adults renal, pulmonary, and osseous lesions are more commonly identified [3].

Most common renal pathologies include angiomyolipomas, cysts, and renal cell carcinoma. An important complication of angiomyolipoma is pseudoaneurysm formation which may result in patients presenting with hematuria. Angiomyolipomas larger than 4 cm and aneurysms larger than 5 mm occurring within these tumors have an increased risk of morbidity [4]. Treatment options maybe nephrectomy or minimally invasive radiological procedures like super-selective coil embolization.

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Case report

A 10-year-old female presented to the Paediatric emergency with complains of hematuria for 5 days and anuria for 1 day. She had right-sided abdominal pain and fever for 2 weeks. There was no history of trauma, surgery, seizures, or any neurodevelopmental delay.

On examination the patient had red papules on her face suggesting adenoma sebaceum (Fig. 1a) and shagreen patch on her back (Fig. 1b). She also had pallor. Her blood pressure was 110/64 mm Hg, heart rate 150 beats per minute, respiratory rate 43 per minute, SpO2 85% and body temperature was 100°F. Blood investigations revealed hemoglobin of 4.2 gm/dL, microcytic hypochromic anemia and elevated leucocyte count.

The emergency ultrasound whole abdomen revealed a large heteroechoic collection in the interpolar region of right kidney (Fig. 2a) with a dependant hyperechoic clot in the urinary bladder.

The initial treatment was with oxygen inhalation, broad spectrum antibiotics and blood transfusion after which she underwent contrast enhanced computed tomography of whole abdomen which revealed a large hematoma (approximately 135 cc) in the interpolar region of right kidney communicating with upper pole calyx (Fig 2c). There was a small homogeneously enhancing exophytic soft tissue lesion without any evidence of fat, suggestive of fat poor angiomyolipoma (Fig. 2b). It also revealed an intensely enhancing pseudoaneurysm within the hematoma arising from a segmental branch of right renal artery. The hematoma was extending into the right ureter causing ureteric obstruction.

Fig. 1 – Images showing adenoma sebaceum (a) on the face including the forehead and shagreen patch on the back (b). (c) The axial NCCT brain image showing calcified subependymal nodule adjoined the frontal horn of left lateral ventricle (arrow). (d) Axial FLAIR MRI image of brain showing hyperintense cortical tubers involving left frontal lobe (yellow arrows).
Fig. 2 – Preoperative images (a) Ultrasound: heteroechoic collection in interpolar region of right kidney. (b and c) Axial CECT: exophytic homogeneously enhancing soft tissue lesion (arrow in b) without identifiable fat, suggestive of fat poor angiomyolipoma. Intensely enhancing pseudoaneurysm (arrow in c) within the right intrarenal hematoma. (d and e) T2W coronal MRI: heterointense hematoma in right kidney and a splenic hamartoma in superior pole of spleen (arrow in e). (f) 3D CT reconstruction showing pseudoaneurysm in right kidney.

and resultant striated nephrogram in the delayed phase with minimal excretion of contrast in right renal calyces. The urinary bladder hematoma measured 50 cc in volume. Magnetic resonance imaging of upper abdomen corroborated with the computed tomography (CT) findings of the right kidney. Additionally, gallbladder calculi (image not shown) and a lobulated exophytic T2 hypointense splenic hamartoma were also noted (Fig. 2d and e).

Based on clinical findings, tuberous sclerosis was suspected, therefore, a CT brain was performed. CT brain revealed a calcified subependymal nodule adjoining the frontal horn of left lateral ventricle (Fig. 1c). Contrast enhanced magnetic resonance imaging brain showed multiple T2 and FLAIR hyperintense cortical tubers involving right posterior parietal, occipital, left frontal lobes (Fig. 1d).

Pediatric surgery department considered nephrectomy to stop the hematuria but this was ruled out as she is a young female and nephrectomy would have led to impaired renal function.

The patient was taken up for selective coil embolization of segmental branches of right renal artery under digital subtraction angiography (DSA) guidance. After right femoral artery access, DSA revealed an aneurysm of size 27 × 17 mm arising from the middle segmental branch of right renal artery (Fig. 3a). Super selective angiography was performed with 2.8F microcatheter. The aneurysm was blocked with microcoils (2 and 3 mm) (Figs. 3b and c). Postcoiling DSA run showed nonopacification of the right renal aneurysm (Fig. 3d).

She was followed up with antibiotic therapy in the first postoperative week and urinary bladder irrigation for clot removal. The postoperative ultrasound revealed a normal renal Doppler (Resistive Index of 0.54 and Pulsatility Index of 0.86) without any features of nontarget embolization. No residual aneurysm was identified on Doppler. A follow-up CT angiography performed on day 7 revealed complete resolution of the pseudoaneurysm (Fig. 3e), normal enhancement of the bilateral renal parenchyma and normal excretion of contrast into the right ureter. There was a significant reduction in the size of the urinary bladder hematoma. The remaining clot was evacuated under cystoscopic guidance and conservative treatment was planned for renal hematoma with serial ultrasound follow-up.

Discussion

Tuberous sclerosis is associated with benign congenital tumors in multiple organs. It has an estimated incidence of 1:6000-12,000 [5]. The classic clinical diagnostic criteria (Vogt triad-seizure, intellectual disability and adenoma sebaceum) are seen in a minority of patients. Therefore, a diagnostic
Fig. 3 - (a-d) Digital subtraction angiography images showing progressive coil embolization by super-selective right renal angiography and blocking of the pseudoaneurysm (d). (e) shows postoperative CT angiography with 3-dimensional reconstruction where the pseudoaneurysm is not visualized.

Angiomyolipomas consist of tortuous and thick-walled blood vessels that lack supportive elastic tissue which results in increased predisposition to spontaneous rupture and formation of intralesional pseudoaneurysm [10]. Thus, the complications of angiomyolipomas are bleeding (which may cause hematuria, intratumoral hemorrhage or retroperitoneal hemorrhage) and mass effects causing abdominal pain and tenderness [11,12].

Benign and asymptomatic angiomyolipomas are managed conservatively. In complications like pseudoaneurysm minimally invasive radiological procedures like super-selective renal embolization can be employed [5,6]. Rarely partial or complete nephrectomy may be needed in uncontrolled bleeding or lack of intervention set-up.

Consent

Obtained from patient’s father.
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