



UNDIAGNOSED CASE OF TUBEROUS SCLEROSIS PRESENTING AS PAIN IN ABDOMEN WITH UNRUPTURED GIANT PSEUDOANEURYSM IN RENAL ANGIOMYOLIPOMA

General Medicine

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ABSTRACT

Tuberous sclerosis complex is a common neurocutaneous syndrome with autosomal dominant inheritance. Angiomyolipoma is benign mesenchymal neoplasm which may be associated with it. Giant pseudoaneurysm in a renal angiomyolipoma associated with tuberous sclerosis complex is rare. The patient may show delayed presentation with pain, abdominal distension or lump. Whenever there are multiple angiomyolipomas in the bilateral kidneys, tuberous sclerosis complex must be ruled out. I report previously undiagnosed case of tuberous sclerosis in a 37year old female who presented with abdominal pain for 5days. There was no past history of seizures, developmental delay, hematuria or flank pain. On clinical examination there was palpable lump in the right lumbar region. The purpose to report this case is to keep high suspicion for tuberous sclerosis in undiagnosed patient with bilateral multiple renal angiomyolipomas which may show delayed presentation with some of its rare complications like pseudoaneurysm as in our case.

KEYWORDS

tuberous, angiomyolipoma, pseudoaneurysm, neurocutaneous.

INTRODUCTION

Tuberous sclerosis complex (TSC) is unusual neuro-cutaneous syndrome with autosomal dominant pattern of inheritance & variable expression. It can affect all ethnic groups, and the estimated frequency is 1 in 6000 to 1 in 10000 live births^(1,2). It may affect virtually every organ system of the body, most commonly the brain, skin, eyes, heart, kidneys and lungs. Angiomyolipoma is benign mesenchymal neoplasm composed of blood vessels, smooth muscle, and varying amounts of fat tissue, and occur in 70-80% of patients with tuberous sclerosis^(1,3,4). I report a case of tuberous sclerosis in a 37years old female.

Case Report

37year old female presented with abdominal pain and distension for 5 days to the emergency department at a tertiary care centre. There was no past history of seizures, developmental delay, hematuria or flank pain. On clinical examination there was non tender palpable lump in the right lumbar region. Ultrasound of abdomen was done which revealed markedly enlarged & heterogenous bilateral kidneys studded with multiple lesions of variable sizes containing echogenic component and vascularity on colour doppler. There was large hypo-to-anechoic area in one of these lesions in right kidney with peripheral eccentric echogenic rim and swirling on colour doppler giving yinyang sign. On Contrast enhanced CT there were multiple renal angiomyolipomas in bilateral kidneys and a giant partially thrombosed unruptured pseudoaneurysm in right kidney in one of the angiomyolipomas (Fig1a). Therefore, for further workup HRCT thorax and non contrast NCCT head was done to look for other lesions, which revealed lymphangiomyomatosis (fig1b) in the bilateral lungs and cortical tubers and calcified subependymal nodules in the brain (fig1c). Retrospectively, patient was examined clinically and was found to have adenoma sebaceum, shagreen patch, hypomelanotic macules and ungual fibroma (fig 2). So based on the imaging and clinical features the diagnosis of tuberous sclerosis was established with unruptured giant renal angiomyolipoma pseudoaneurysm.

pseudoaneurysm (arrow) in one of the angiomyolipoma in right kidney with eccentric thrombus (arrowhead). Fig 1b is the CT thorax axial image showing multiple thin walled air containing cysts in bilateral lungs suggestive of lymphangiomyomatosis. Fig 1c is the NCCT head axial image showing the calcified subependymal nodules (arrow). Fig 2 shows the adenoma sebaceum on face.

DISCUSSION

TSC-associated angiomyolipoma are multiple, large in size and more likely to cause spontaneous haemorrhage than the sporadic forms⁽⁵⁾. There is increased incidence of pseudoaneurysm formation in angiomyolipomas larger than 4cm and increased risk of rupture if the pseudoaneurysm is 5 mm or large⁽⁶⁾. Few cases of large pseudoaneurysm associated with TSC are described in the literature^(7,8). Tuberous sclerosis may have varied presentations at time of diagnosis like seizures, skin lesions or angiomyolipomas in kidneys on sonography, rarely it may present as abdominal lump with aneurysm as in our case which may need further imaging/clinical evaluation to rule out tuberous sclerosis complex.

REFERENCES:

- Northrup H, Krueger DA; International Tuberous Sclerosis Complex Consensus Group. Tuberous sclerosis complex diagnostic criteria update: recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. *Pediatr Neurol*. 2013;49:243-54. 2.
- Umeoka S, Koyama T, Miki Y, et al. Pictorial review of tuberous sclerosis in various organs. *Radiographics*. 2008;28:e32.
- Manoukian SB, Kowal DJ. Comprehensive imaging manifestations of tuberous sclerosis. *AJR Am J Roentgenol*. 2015;204:933-43. 4.
- Ranke FM, Faria IM, Zanetti G, et al. Imaging of tuberous sclerosis complex: a pictorial review. *Radiol Bras*. 2017 Jan/Fev;50(1):48-54.
- Nelson CP, Sanda MG (2002) Contemporary diagnosis and management of renal angiomyolipoma. *Urology* 168: 1315-1325.
- Yamakado K, Tanaka N, Nakagawa T, Kobayashi S, Yanagawa M, et al. (2002) Renal Angiomyolipoma. *J Urol: Relationships between Tumor size, Aneurysm Formation, and Rupture. Radiology* 225: 78-82.
- Choh NA, Choh SA, Yousuf R, Jehangir M (2009) A giant aneurysm arising from renal angiomyolipoma in tuberous sclerosis. *Arch Dis Child* 94: 47-48.
- Lapeyre M, Correa JM, Ortonne N, Balleyguier C, Helenon O (2002) Color-Flow Doppler Sonography of Pseudoaneurysms in Patients with Bleeding Renal Angiomyolipoma. *AJR Am Roentgenol* 179: 145-147.

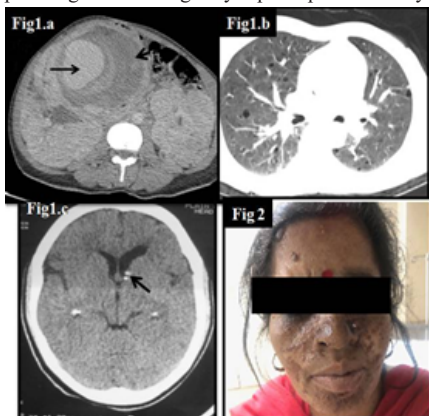


Fig1a is the CECTabdomen axial image which shows the giant