

Case Report

Tuberous sclerosis with giant renal angiomyolipoma and sclerotic skeletal lesions in a geriatric patient: role of imaging

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ABSTRACT

Tuberous sclerosis is a rare inherited neurocutaneous syndrome, which has multisystem involvement. We present a geriatric female patient with chronic abdominal pain who was diagnosed with giant renal angiomyolipoma (AML) and multiple sclerotic skeletal lesions. Radiological imaging played an important role in identifying these two features and diagnosing tuberous sclerosis in our case.

Keywords: Angiomyolipoma, Computed tomography, Sclerotic lesions, Tuberous sclerosis, Ultrasound

INTRODUCTION

Tuberous sclerosis complex is a rare neurocutaneous syndrome with an autosomal dominant inheritance. It is characterized by benign congenital neoplasms involving various organs and systems. It has several major and minor physical and radiological features which are helpful in arriving at diagnosis. We present a geriatric female patient with renal angiomyolipoma (AML) and sclerotic bone lesions, without cutaneous and neurological involvement. We also emphasize the importance of imaging in arriving at the diagnosis in our case.

CASE REPORT

A 76 year old female patient presented with chronic abdominal pain and palpable mass per abdomen in left lumbar region. She had these complaints since 5 to 6 months which had aggravated recently since 1 week. She had no past history of hospital admissions. Her obstetric history was unremarkable with 3 normal vaginal deliveries at home and she had attained menopause 30 years back. There were no cutaneous lesions or

neurological symptoms. Her routine laboratory investigations were within normal limits including renal function tests. She was referred for ultrasound and contrast computed tomography to our department.

Ultrasound (Figure 1) revealed a fairly defined heterogeneous hyperechoic large mass lesion arising from interpolar region of left kidney extending exophytically into perinephric spaces. Doppler interrogation showed multiple vascular channels within.

Plain and contrast enhanced CT abdomen (Figure 2 and 3) revealed a well circumscribed fat attenuation mass lesion arising from interpolar region of left kidney, measuring ~ 12.7 x 11.5 x 10.8 cm (antero-posterior x transverse x cranio-caudal) in maximum dimensions. Multifocal minimally enhancing soft tissue attenuation areas were noted within. There was no evidence of calcification. Few prominent vascular channels were noted within the lesion, without evidence of aneurysm / arterio-venous malformations. The lesion was noted to extend exophytically into the anterior perinephric space upto midline, causing splaying of the left renal vessels

and renal pelvis with significant mass effect and narrowing of pelvi-ureteric junction.

There was secondary dilatation of upper calyceal system; however normal enhancement and excretion of contrast from remnant left kidney was noted (Figure 4). There was anterior displacement of adjacent bowel loops by the lesion. Mild extension into posterior perinephric space was also present

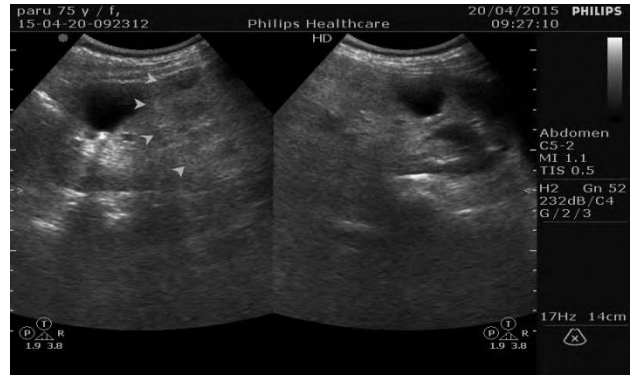


Figure 1: Ultrasound through left kidney showing fairly defined heterogeneous hyperechoic lesion involving interpolar region of kidney.



Figure 2: Plain (A) and contrast enhanced (B) axial computed tomography at the level of kidneys, showing large well circumscribed fat attenuation lesion in interpolar region of left kidney with perinephric extension causing splaying of renal pelvis.



Figure 3: Sagittal (A) and coronal (B) CT sections showing the lesion.

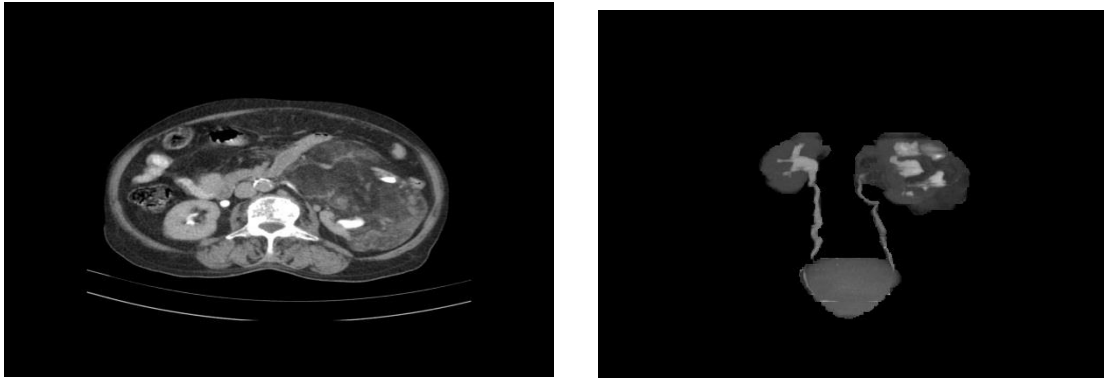


Figure 4: Delayed phase axial CT (A) and maximum intensity projection (MIP) images (B) showing excretion of contrast.

Craniocaudally the lesion extended from L1-L2 to L4-L5 intervertebral disc level. There were few prominent left renal hilar lymph nodes. Bone window (Figure 5) revealed multiple fairly defined sclerotic lesions in visualized spine, bilateral ribs and pelvis. There were mild bilateral pleural effusions. No focal pulmonary parenchymal lesions were noted. CT brain was performed to rule out subependymal nodules and astrocytomas, which was normal.

Echocardiography was performed to look for rhabdomyomas, which was unremarkable except for mild LVH.

Based on imaging findings of fat attenuating lesion of kidney, likely representing angiomyolipoma and multiple sclerotic lesions in skeletal system, a probable diagnosis of tuberous sclerosis was made without cardiac or neurological involvement.

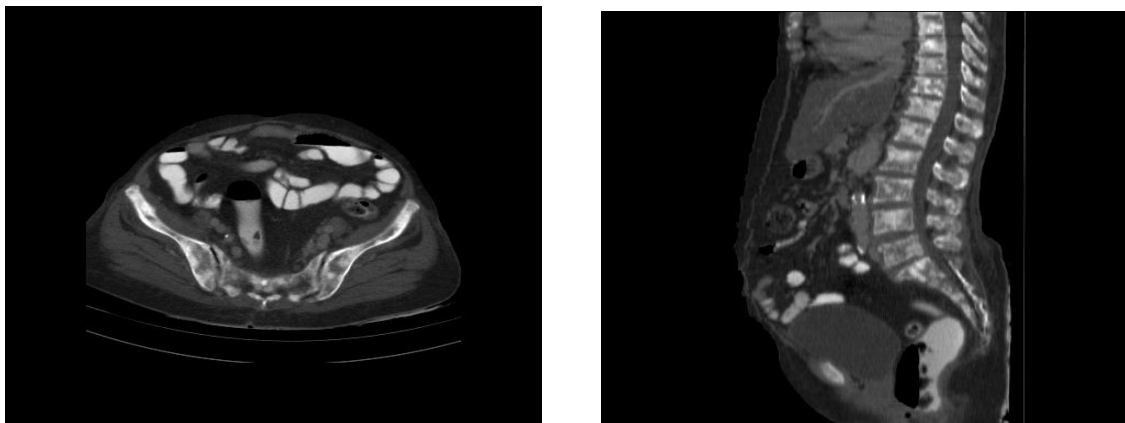


Figure 5: Axial (A) and sagittal (B) CT sections in bone window showing multiple fairly defined sclerotic lesions involving pelvic bones and thoraco-lumbar spine.

She underwent left radical nephrectomy (Figure 6) and histopathology of the lesion confirmed the diagnosis of angiomyolipoma.

Post operatively the patient developed aspiration and required ventilator support for 5 days. However patient could not recover and died on 5th post-operative day.



Figure 6: Gross specimen of the lesion showing fatty components.

DISCUSSION

Tuberous sclerosis is a neurocutaneous syndrome characterized by hamartomatous lesions involving various organ systems. It is known to be caused by mutations involving TSC1 (Chromosome 9) and TSC2 genes (chromosome 16). The classic clinical triad comprising of epilepsy, adenoma sebaceum and mental retardation occurs rarely. The diagnosis is based on various major and minor criteria applied to physical and radiological findings. Hence identification of the components of this syndrome radiologically plays an important role in diagnosis.¹

Cutaneous involvement may be in the form of hypopigmented macules, shagreen patches, facial angiofibromas (adenoma sebaceum) and ungual fibromas.¹ Neurological manifestations include white matter abnormalities (radial migration lines), cortical tubers, subependymal nodules, subependymal giant cell astrocytomas (SGCAs) and microcephaly.^{1,2} Cardiac rhabdomyomas and pulmonary lymphangiomyomatosis (LAM) are thoracic manifestations. Renal involvement can occur in the form of multiple renal cysts, renal AML and renal cell carcinoma.^{1,3,4,5} Skeletal involvement with bone cysts, sclerotic lesions, periosteal new bone formation and scoliosis may be noted.¹ Miscellaneous rare manifestations include retroperitoneal LAM, hepatic hamartomas & AMLs and intestinal polyposis.¹

The major features include facial angiofibromas, hypomelanotic macules, cortical & subependymal tubers, subependymal giant cell astrocytomas, renal AMLs, cardiac rhabdomyoma, pulmonary LAM and retinal astrocytomas. All the other manifestations explained before are the minor features of the disease. A 'definite' diagnosis of TS is made with 2 major features or 1 major with 2 minor features. A 'probable' diagnosis is with 1 major and 1 minor feature and a 'possible' diagnosis is made with 1 major or 2 or more minor features.¹

Our case had 1 major feature (renal AML) and 1 minor feature (sclerotic skeletal lesions) leading to a 'probable' diagnosis of tuberous sclerosis.

AMLs are benign renal neoplasms and as the name suggests are composed of vascular, smooth muscle and fat components. Its prevalence ranges from 0.3 to 3% with peak age at presentation being 43 years.⁶ They may occur in an isolated form or may have syndromic association.⁶ Cases associated with tuberous sclerosis may have multiple AMLs or giant AMLs. The blood vessels within the lesion are abnormal without internal elastic lamina and replacement of smooth muscle with fibrous tissue, predisposing to aneurysm formation with the risk of rupture and hemorrhage leading to hypovolemic shock.³

Ultrasound reveals a well demarcated hyperechoic lesion with posterior shadowing.⁶⁻⁸ Vascular components show colour flow on Doppler interrogation. Computed tomography is more sensitive in identifying macroscopic fat within these lesions, which is pathognomonic. Contrast administration helps in identifying vascular components and also to look for aneurysm formation and rupture.^{8,9} CT also better delineates the extension of the lesion into perinephric space and its mass effect on adjacent structures. MRI also helps in detection of fat, which appears hyperintense on both T1 and T2 FSE images with suppression on fat saturated sequences. Perinephric extension is equally well demonstrated on MRI.^{1,6,9}

Sclerotic skeletal lesions are one of the minor criteria, which may involve axial skeleton. The abnormal sclerotic bone islands are usually multiple and may be asymptomatic, detected incidentally when imaging is done for other complications of the disease.¹

CONCLUSION

Tuberous sclerosis is a rare neurocutaneous disorder with multisystemic involvement and requires various major and minor criteria for diagnosis. Imaging plays an important role in identifying various components of tuberous sclerosis and arriving at a diagnosis. Upon identification of one major criterion, a thorough search for other major and minor features is necessary to diagnose and thus avoid various complications arising out of it.

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REFERENCES

1. Umeoka S, Koyama T, Miki Y, Akai M, Tsutsui K, Togashi K. Pictorial review of Tuberous Sclerosis in various organs. *Radiographics.* 2008;28(7):e32.
2. Kalantari BN, Salamon N. Neuroimaging of Tuberous Sclerosis: Spectrum of pathologic findings and frontiers in imaging. *AJR* 2008;190:W304-9
3. Radhakrishnan R, Verma S. Clinically Relevant Imaging in Tuberous Sclerosis. *J Clin Imaging Sci.* 2011;1:39.
4. Logue LG, Acker RE, Sienk AE. Angiomyolipomas in Tuberous Sclerosis. *RadioGraphics.* 2003;23:241-6
5. Casper KA, Donnelly LF, Chen B, Bissler JJ. Tuberous Sclerosis Complex: Renal imaging findings. *Radiology.* 2002;225:451-6
6. Kotis A, Lisgos F, Karatapanis S. Ultrasound and CT imaging assessment of renal angiomyolipoma. *BMJ Case Reports.* 2010;10:1136.
7. Maizlin ZV, Gottlieb P, Corat-Simon Y, Strauss S. Various appearances of multiple angiomyolipomas

in the same kidney in a patient without Tuberous Sclerosis. *J Ultrasound Med.* 2002;21:211-3.

8. Prando A. Radiological classification of renal angiomyolipomas based on 127 tumors. *Int Braz J Urol.* 2003;29:208-16
9. Bosniak MA, Megibow AJ, Hulnick DH, Horil S, Raghavendra BN. CT diagnosis of renal

Angiomyolipoma: The importance of detecting small amounts of fat. *AJR Sept.* 1988;151:497-501.

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