

A Rare Case Report of Angiomyolipoma Kidney Associated with Tuberous Sclerosis

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ABSTRACT : Angiomyolipomas are rare, benign tumours which are composed of an intimate admixture of blood vessels, smooth muscle cells and fat in varying proportions and hence the name. They occur at many sites, more commonly in the kidney. Usually they present as an incidental finding or with retroperitoneal haemorrhage in adults. They are seen in 25-50% of the patients with tuberous sclerosis.

KEY WORDS: Angiomyolipoma, kidney tumours, tuberous sclerosis.

I. INTRODUCTION

Angiomyolipoma (AML) is an uncommon, benign, mesenchymal tumour (2-6.4% of all the kidney tumours) which arises from either the renal pelvis or the sinus, which is composed of an intimate admixture of blood vessels, smooth muscle cells and fat, hence the name¹. In 5% of the tumours, fatty elements can be detected only at microscopy². AML commonly occurs in women who are of the age group of 40- 70 yrs¹. It may occur sporadically as isolated lesions (80-90%) or in association with an autosomal dominant disorder, tuberous sclerosis (TSC) (25-50%). Sporadic angiomyolipomas are usually unilateral and focal, whereas those which are associated with TSC are bilateral and multifocal and can occur at any age and in either sex. Usually it is asymptomatic and is often found incidentally in USG or CT. Sometimes it may present with retroperitoneal haemorrhage in adults. Now it is recognised as they belong to the family of perivascular epithelioid cell tumours (PEComas). Angiomyolipoma is benign and the transformation to sarcoma from conventional AML is very rare³.

II. CASE SUMMARY

A 19 year old female presented with a mass in the right loin with a dragging pain, with clinical features suggestive of tuberous sclerosis. CT scan (Fig.1,2) showed - Right kidney is enlarged in size, lobulated in outline. An irregular lobulated large heterogeneous space occupying lesion is noted arising from lower pole. The space occupying lesion is solid and mostly contains fatty components, dilated tortuous vessels and soft tissue. A rounded cystic component also noted. The lesion measures 13.8x12.5 cm in anteroposterior and transverse axes respectively. SOL extends upto midline. Upper and midpole of right kidney also contains multiple small fat density SOLs. Pelvi-calyceal system is not dilated. No calculus is seen. Left kidney and liver scan showed multiple small fat containing lesions. CT guided FNAC was done and suggestive report of angiomyolipoma was made. Right sided nephrectomy along with contralateral kidney and liver biopsy was done. Microscopy of rt. renal tumour revealed the classical features of angiomyolipoma, which comprised of mature adipose tissue, thick walled blood vessels and bundles of smooth muscle cells. No epithelioid component was seen. Contralateral kidney biopsy showed the same features of classical angiomyolipoma. However, the liver biopsy showed only mature adipose tissue. Neither angiomatous component nor muscular element was noted.

Histopathological findings

Gross examination revealed the rt. renal tumour weighing 50 gms and measured 10x7x4cms in diameter. The external surface of the tumour was yellowish to grey brown, with a lobulated surface. The capsule was intact and it could be easily stripped off (Fig.3). The cut section was greasy and yellowish to grey brown in colour. The mass arose from the renal pelvis and it was soft to firm in consistency. Blood oozed out on cutting the fresh specimen. The normal part of the kidney was also identified in the specimen (Fig.4).

The microscopic examination of rt.renal tumour mass revealed mature adipose tissue, thick walled blood vessels and bundles of smooth muscle cells, which are classical features of angiomyolipoma. A part of the normal kidney was seen adjacent to the tumour. No epithelioid component was seen. There was no evidence of vascular invasion and it was not associated with epithelial cysts and renal cell carcinoma(Fig.5,6,7). M/E of Contralateral kidney biopsy showed the same features of classical angiomyolipoma.

The liver biopsy revealed only mature adipose tissue.Neither angiomatous component nor muscular element was noted.

Immunohistochemistry showed HMB-45 positivity(Fig.8).

III. DISCUSSION

Renal angiomyolipoma have an incidence of 0.3–3% and arise from the mesenchymal elements of the kidney.Angiomyolipomata may occur as an isolated phenomenon or as part of the syndrome associated with tuberous sclerosis. The overall female:male predominance is approximately 4:1 which is suggestive of a hormonal component to tumour growth². Isolated angiomyolipomas occur sporadically, account for 80% of angiomyolipomas, are usually solitary and occur almost exclusively in women in the fourth to fifth decade of life (mean age 43)⁴. Angiomyolipomata associated with tuberous sclerosis are typically larger than isolated angiomyolipomata, have multifocal or bilateral disease, tend to occur in younger patients (third decade), and require careful screening for the presence of renal tumours⁵. With tuberous sclerosis associated angiomyolipomata the male:female distribution is nearly equal; however women outnumber men in terms of prevalence.They are not encapsulated, but well demarcated and may be locally infiltrative and cut surface may resemble that of a lipoma.

AML occurs not only as a rare tumour which is restricted to the kidney, but also as a biologically fascinating and morphologically heterogeneous entity. It is also seen at various other sites like the skin, appendix, colon, liver, lung and the smooth muscle fibres. Most small angiomyolipomata are asymptomatic and found incidentally on imaging studies, usually incidental from US or CT scans performed for unrelated clinical indications. They can vary in size from a few millimeters to larger than 20 cm. In the minority of patients that are symptomatic, they classically present with flank pain (53%), a palpable tender mass (47%) and gross haematuria (23%); this is known as 'Lenk's triad'⁶. Other symptoms associated but noted less frequently include nausea or vomiting, fever, anaemia and blood pressure changes⁷.Histology of the tumour varies depending on relative proportion of fat,smooth muscle and blood vessel⁸. Radial arrays of smooth muscle fibres around the blood vessels are frequently noted.Smooth muscles are also seen in bundles and also as individual fibres.The cells are spindle shaped but occasionally polygonal and rounded showing an epithelioid appearance with abundant eosinophilic cytoplasm.Blood vessels are thick walled like that of arteries.Nuclear pleomorphism and mitotic figures may be present but does not contribute to adverse prognosis.Local lymph nodal involvement may be present^{9,10,11}. but this does not signify metastasis.

Histologically, it can mimic renal cell carcinoma and it can be diagnosed accurately by immunostaining with HMB-45, Melan-A, CD68, CD117 and Ki-67¹². The importance of HMB-45 Immunoreaction depends on monoclonal antibody reaction specifically with premelanosomes in the smooth muscle cells. No other benign or malignant renal tumors show staining with HMB-45. This confirmation is important for the correct definition of the nature of the tumor and the most appropriate procedures for the patient^{14,15}. It mimics the same condition in imaging studies when it contains less fat.Conventional AML has got a very good prognosis as compared to the rare epithelioid variant of AML, which is potentially malignant¹³.D/d includes lipoma if it contains predominantly fat,leiomyoma if muscle ,epithelial tumours of kidney if epithelioid features.

In our case,the patient is a 19 year young female c/o dragging pain over rt. lumbar region,rt. lumbar mass,no haematuria and presented with small fibrous papule(adenoma sebaceum)in bilateral distribution over the face(Fig.9) and shagreen patch over the back(Fig.10) which are suggestive of TS The angiomyolipoma was bilateral involving both kidneys –rt. more than left .The liver biopsy showed only adipose tissue which may be due to involvement of liver by same pathology.These features are in keeping with multifocal involvement as seen in tuberous sclerosis.On histology ,our case showed presence of fat,muscle and blood vessels in both kidney specimens.So diagnosis was not difficult.The biopsy from the liver revealed only fat which may be present as a part of AML as discussed above.

IV. CONCLUSION

Renal angiomyolipoma is an uncommon benign tumor, which may possess a challenge for clinical and pathological diagnosis. The presence of fat is highly suggestive of classic AML by CT; epithelioid AML contains no fat in CT. AMLs are characteristically positive for HMB-45 . USG, CT scan FNAC, histopathology

and immunohistochemistry play a vital role in the diagnosis of renal angiomyolipoma. Presence of bilateral or multifocal tumours are strongly in favour of tuberous sclerosis. So, we should always keep in mind the association between the two entities.

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Figure 1. CT scan showing kidney lesions.

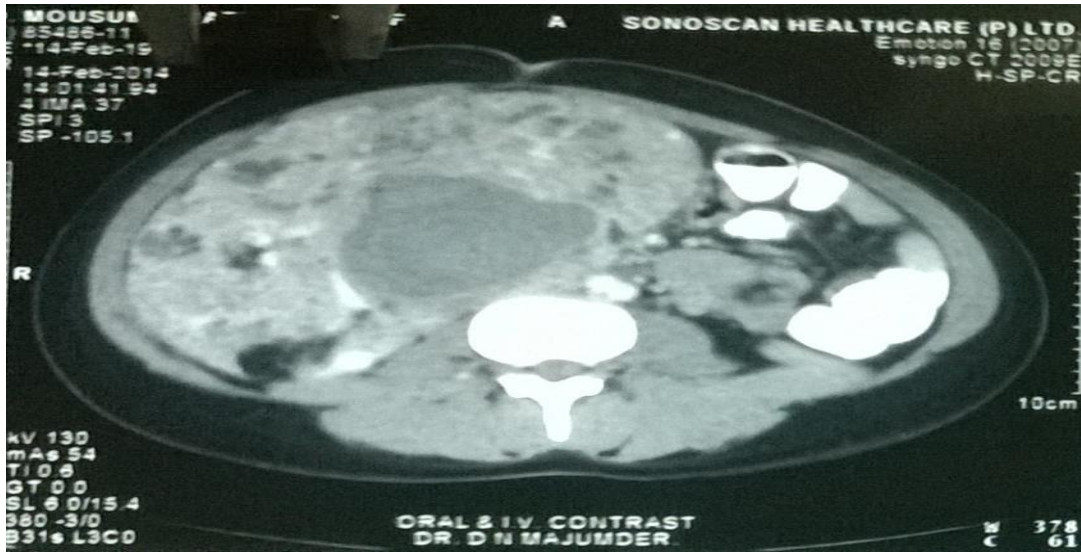


Figure 2.CT scan highlighting the variegated appearance of the tumour mass.

Figure 3.Gross picture of renal tumour



Figure 4.Gross specimen ,the cut surface highlighting the renal origin

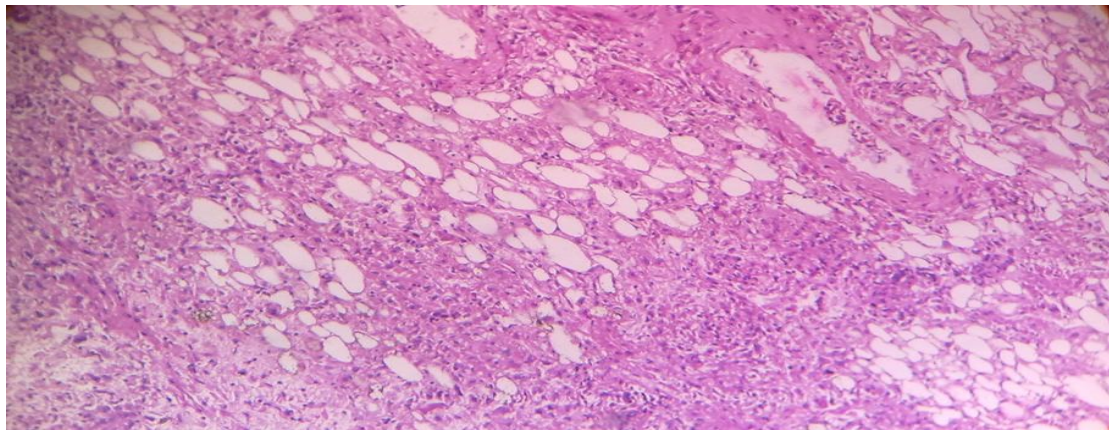


Figure 5.L/P view .tumour composed of fat and blood vessels and smooth muscles..

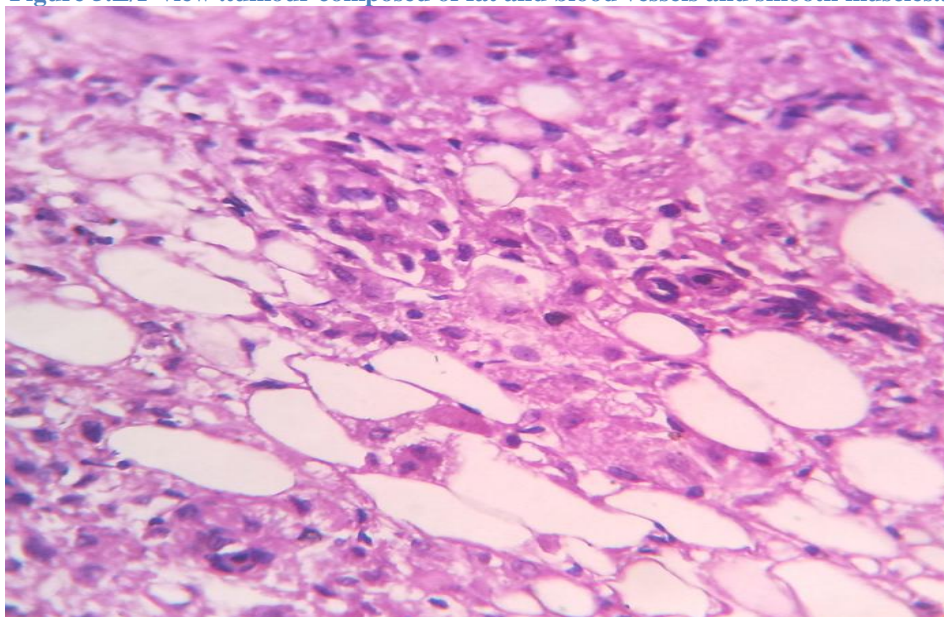


Figure 6.H/P view .tumour composed of fat and blood vessels.

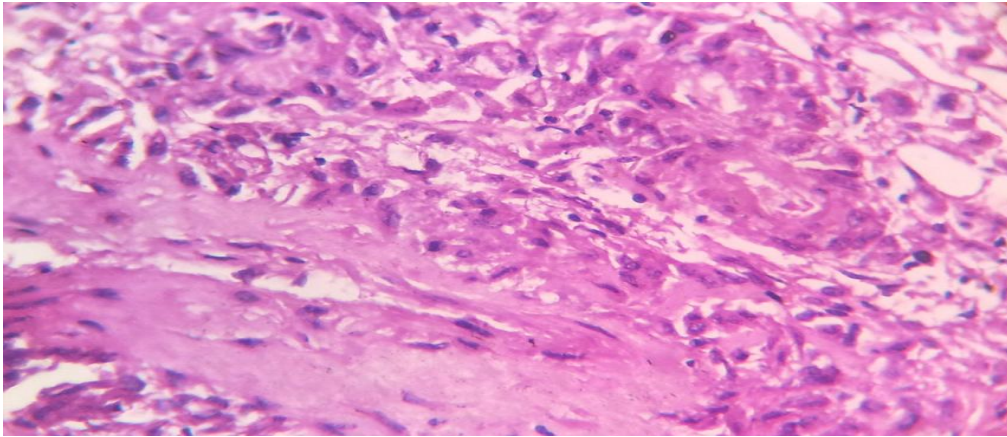


Figure 7.H/P view .Thick walled blood vessel with radially arranged smooth muscle fibres.

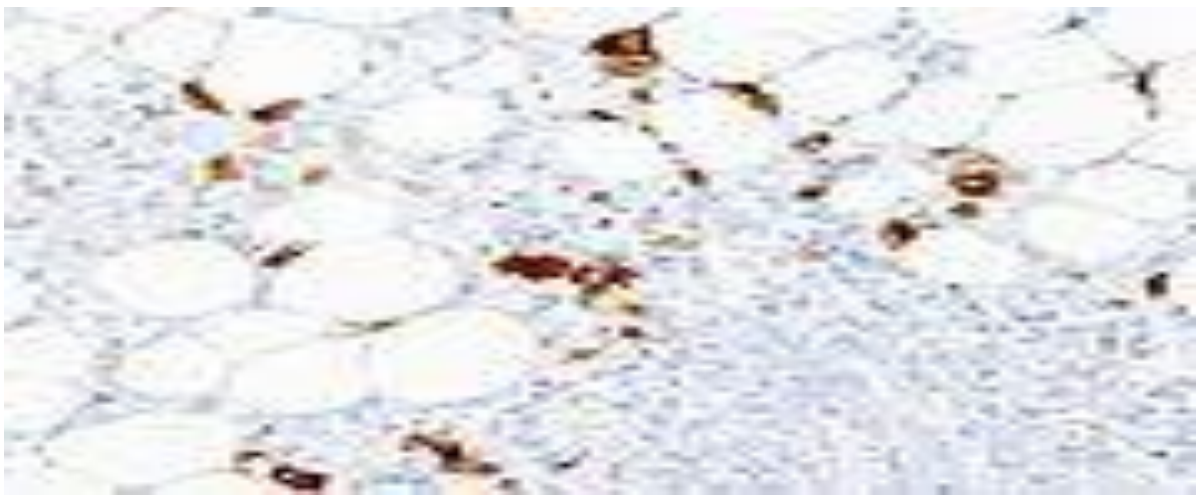


Figure8.IHC showing HMB45 positivity

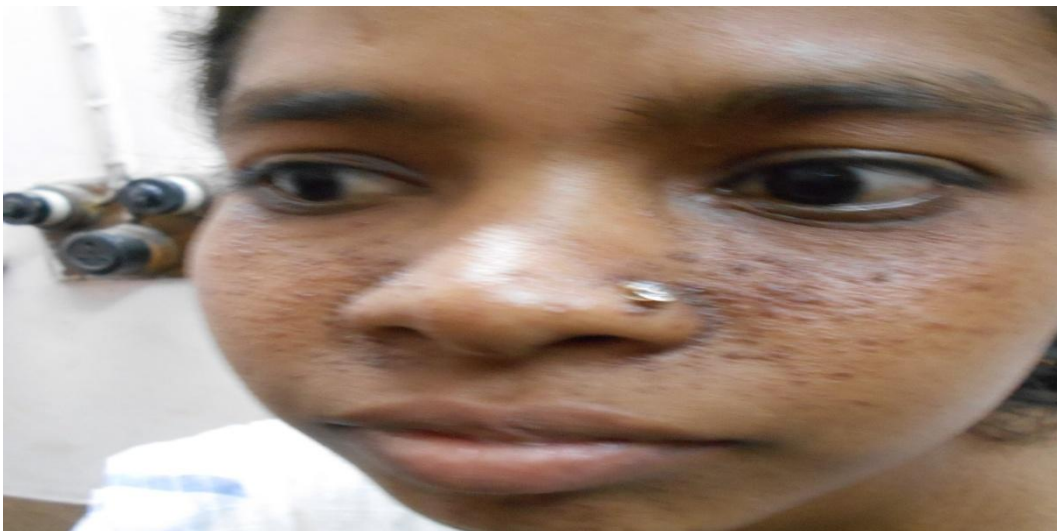


Figure 9.Bilateral distribution of adenoma sebaceum



Fig.10.Shagreen patch over the back