Atypical Renal Lesions in Tuberous Sclerosis: A Case Report

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A case of hypoechoeic and enhancing multiple renal lesions observed in 35-year-old male is reported. The patient was a known case of seizure disorder which was poorly controlled by medication. After Magnetic resonance imaging study of the brain patients was found to be having multiple subependymal lesions consistent with Tuberous sclerosis.
The patient was investigated further with ultrasonography and Computerized scanning of the abdomen which revealed multiple bilateral renal lesions which were though not typically consisted with the common entity that is angiomyolipoma except one lesion which was consistent with the features of angiomyolipoma. All the lesions were hypoechoeic in the ultrasonography and were enhancing in the contrast Computerized scanning.
The largest lesion was 38mm and renal function was normal biochemically. The patient was planned for the exploration of the kidney for the possible partial nephrectomy. As this tumor had none of the typical elements of classic AML, the final pathological diagnosis would have proved the type of lesion in this patient of tuberous sclerosis.
Keywords: adenoma sebaceum, angiomyolipoma, hypoechoeic, von Hippel-Lindau syndrome.

Tuberous sclerosis is transmitted as an autosomal dominant trait. It is characterized by the formation of angiomyolipomas or tubers in the skin (called adenoma sebaceum), brain, kidneys, and other organs. Two different genetic loci have been identified: one on chromosome 9 and one on chromosome 16. A classic triad of seizure, mental retardation & adenoma sebaceum is observed in less than ten percent of the patient diagnosed with tuberous sclerosis.
The most common renal manifestation of TS is the formation of angiomyolipomas, benign cysts and, much less often, lymphangiomas also can occur. The angiomyolipomas are composed of varying amounts of mature adipose tissue, smooth

muscle, and thick-walled blood vessels. Patients with bilateral renal angiomyolipomas have an 80 to 90 percent chance of having tuberous sclerosis, whereas those with a single lesion may not have systemic disease.[1]

Many patients with TS have no symptoms referable to the kidney however, in very few patients they might present with the history of hematuria or flank pain, although most patients had angiomyolipomas on ultrasonography. Renin-dependent hypertension, due to focal areas of ischemia around lesions, and chronic renal failure also can occur, simulating the clinical picture of autosomal dominant polycystic kidney disease. Renal cell carcinoma is another potential complication of tuberous sclerosis. It appears to occur in 1 to 2 percent of patients, which represents a substantially lower frequency that seen in von Hippel-Lindau syndrome.[2]

The widespread use of abdominal ultrasonography has increased the detection of incidental findings of renal angiomyolipomas. The primary indications for intervention are pain refractory to medications, bleeding, and suspicion of malignancy.

Case Report

A 35-year-old male presented with 15 years history of seizure disorder and it was a week ago prior to presentation he had last seizure despite being on regular antiepileptic medication. He neither has any symptoms related to bladder or bowel systems nor did any symptoms relate to chest system however, he had noticed some rough skin lesions on his face for few years. He denied any history of abnormal lumps in the body and there were not similar history including any kidney related problems in his entire family.

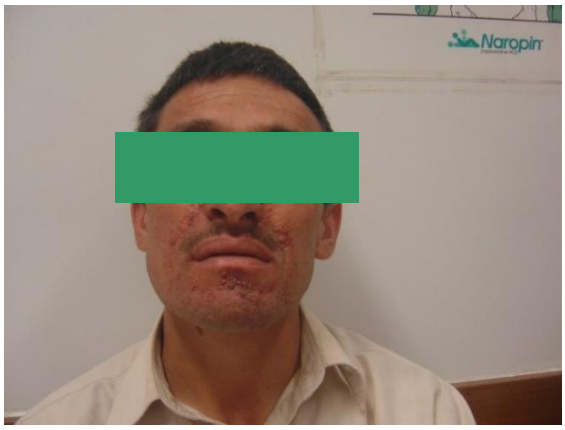

Figure 1: Papulo-nodular lesions over the cheek area suggestive of adenoma sebaceum

On examination he was of average intelligence with normal physics, normotensive and vitally stable. There were papulo-nodular lesions over his malar and cheek region with hyperemia about which he was not bothered **(Figure 1)**. Abdominal and digital rectal examinations were unremarkable and genitalia were normal.

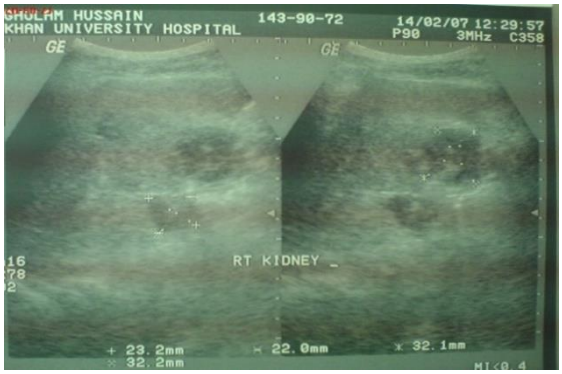

Figure 2: USG abdomen showing hypoechoeic multiple renal lesions

With these findings patients was further investigated in our institute considering his uncontrolled seizure. MRI of the Brain was performed and which revealed multiple calcified subependymal nodules with

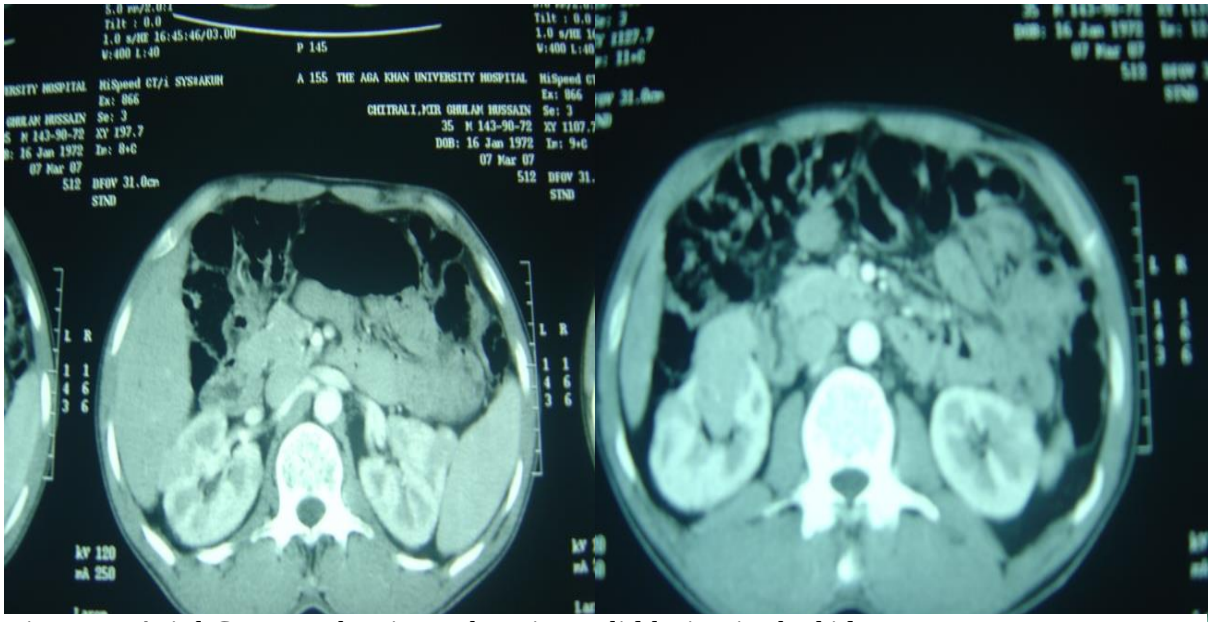

Figure 3: Axial CT scan showing enhancing solid lesion in the kidney

multiple cortical abnormalities consistent with tuberous sclerosis. After that we investigated further with simple ultrasound of the KUB which revealed multiple hypoechoeic lesions in the right kidney with the maximum size was 34 mm in the largest dimension which were relatively avascular along with one cortical cyst of 14 mm and in its largest dimension **(Figure 2)**.

There was only one small lesion measuring about 8mm was consistent with angiomyolipoma according to its echogenic pattern. Renal functions and Urine microscopy examination were normal. Ultrasonography picture is shown in the slide below. Finally we planned to get a CT- KUB done on him and which revealed bilateral enhancing solid lesions consistent with Tuberous sclerosis **(Figure 3)**. There were about five such lesions in the right kidney with largest one measuring about 38mm in its largest dimension. There was small solid enhancing lesion in the left kidney as well which was not picked up by the ultrasonography.

CT picture is shown below.In this patient after serial of investigations most of the lesions were not consistent with the typical angiomyolipomas though one lesion in the ultrasound imaging was suspicious according to it echogenecity. Typically angiomyolipomas have the density similar to the density that of adipose tissue in the radiological imaging however, depending upon the amount of adipose tissue present in the lesions its density varies. Angiomyolipoma with a less amount of fat makes a diagnosis very difficult radiologically.

Patient was planned for the exploration of right kidney and possible partial nephrectomy owing to the size of the lesion the largest one being 38mm, after thorough work up and discussion. He was explained thoroughly about the procedure including important of regular follow up study however he refused to undergo despite being explained about the possible dreadful consequences.

Discussion

Renal manifestation used to be the second

commonest cause for the mortality after central nervous system complications in the patients with tuberous sclerosis. However, with an advent of good medications for the control of seizure, renal complications is said to be the leading cause of mortality in them. Apart from the clear indications for the intervention like refractory to medications, bleeding, and suspicion of malignancy, intervention in asymptomatic patients is debatable. Various studies have come up with different recommendation however the size of the lesions being the most important factor predicting intervention.

A study conducted by Harabayashi et al reported that intervention was required in 20 percent of tumors less than 4 cm in diameter, 50 percent of tumors 4 to 10 cm, and 100 percent of tumors 10 cm or greater. Among 14 tumors followed by observation, eight grew and four required intervention.[3]

A study done by Van Baal JG et al concluded that cysts greater than 3.5 to 4 cm in diameter are more likely to increase in size and to produce symptoms requiring intervention.[4]

In a review of Lemaitre L et al angiomyolipomas ≥4 cm, 82 to 94 percent were symptomatic and 50 to 60 percent bled spontaneously.[5]

Dickinson M study recommended that the symptomatic lesions less than 4 cm in diameter can be monitored with periodic ultrasonography. Potential indications for intervention include progressive growth of the mass or symptoms (such as flank pain, hematuria, or a palpable mass), which are commonly present with angiomyolipomas larger than 4 cm.[6]

Conclusion

Renal sparing approaches are preferred when intervention is required because of its bilateral in character most of the times. This includes partial or nephron-sparing surgeries when there is associated with a risk of significant hemorrhage or transcatheter embolization which is mostly preferred modality indeed.[7] Embolization reduces the risk of hemorrhage by destroying the blood supply to the angiomyolipoma. Total Nephrectomy may be indicated in case of a nonfunctioning kidney causing uncontrolled hypertension, local tissue invasion, tumor in the renal vein, and very strong evidence of malignancy.[8]

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