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CASE REPORT

Rare Case of Giant Bilateral Renal Angiomyolipoma

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ABSTRACT

Renal hamartoma, commonly known as renal angiomyolipoma (AML), is an uncommon solid tumor that does not exhibit malignant traits. Renal AML is inherited via an autosomal dominant pattern. We present a case of 25 years old female who had bilateral renal giant AML's. Bleeding is a major risk if they are big enough and this requires urgent embolization if needed. Conservative treatment is not recommended in such cases and mostly nephrectomy (partial or total) is treatment of choice considering high bleeding risk in such giant AML's. Giant AML's with size more than 20 cm are rarely reported till date in Pakistan.

Key-words: Angiomyolipoma, Renal AML, Tuberous Sclerosis, Radiology, Pakistan.

Authors' Contribution:
[1,2]Conception; Literature research; manuscript design and drafting; [2,3]Critical analysis and manuscript review; [1,3]Data analysis; Manuscript Editing.

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Introduction

Tuberous sclerosis complex (TSC) is a rare genetic disesase with autosomal domiant pattren of inheritance.[1] The characterizing feature of this disease is formation of bening tumours called hamartoms in multiorgans of body including brain, eyes, heart, lungs, kidneys and skin leading to broad range symptomps and complications such as seizures, developmental delays, intellectual disabilities, skin abnormalities and kidney problems.[2] The cutanoeus and renal systems are predominantly affected. The commonest renal manifestations are angiomyolipomas and renal cysts.[3]

Renal angiomyolipomas (RAML) can occur sporadically or in association with TSC. Those assocaited with TSC tends to be large and multifocal and have greater tendency for retropeitoneal hemorrhage.[4] Histologically RAML consists of blood vessels, smooth muscle, and adipose tissue in differing ratios.[5] Renal AMLs that exceed 10 cm in size are classified as 'giant' AMLs and occurrence of these are sparsely reported in literature.[6] The enlarging AMLs have increased incidence of compression syspmtoms and greater propensity to bleed because of development of hemorrhagic aneurysms in them.[7] We hereby report a rare case of bilateral giant AMLs associated with TSC-complex, focussing on imaging findings.

Case Presentation: 25 years old female presented with active complains of abdominal discomfort for the last 2 weeks. Historically she has had cutaneous manifestations and was also treated for seizures which were now stable and for which she does not use medication. Patient was referred to radiology

department for CT scan. CT scan renal dynamic was performed according to departmental protocols. Bilateral kidneys were replaced by large differential density masses extending till bilateral hemipelvis; having predominantly fatty component with CT density of approximately -30 to -70 HU. Soft tissue and vascular component of these masses showed heterogenous arterial and venous phase enhancement with washout on delayed phase. No definite internal foci of calcification was seen. The right-sided mass measured approximately 13 x 10 x 28.3 cm and the left-sided mass measured approximately 10 x 8.5 x 22.7 cm in maximum AP X TR X CC dimensions.

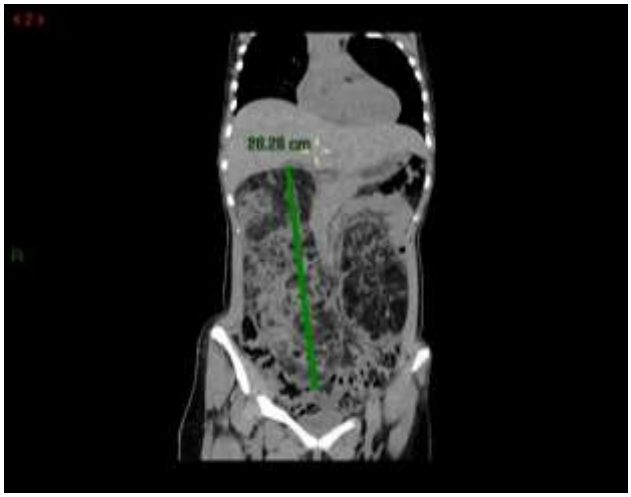

Figure 1: Right sided AML.

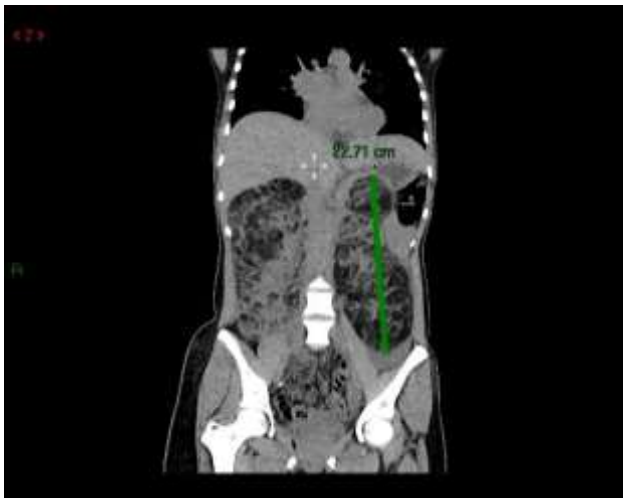

Figure 2: Left sided AML.

Grossly abnormal intrarenal arteries that were beaded with multiple small aneurysms. Normal excretion of the contrast medium was seen on both sides. Mild left and minimal right perinephic free fluid was noted along lower poles with CT attenuation of approximately 20-25HU. No definite intrarenal, perinephric hemorrhage or hemoperitoneum was seen in current scan. Mass effect on adjacent structures displacing bowel loops anteriorly was noted.

Findings were consistent with giant AML keeping in view patient known tuberous sclerosis history.

Liver was normal without focal lesion, biliary ductal dilatation or vascular thrombosis. Spleen, pancreas and gallbladder appear normal. Adrenals were compressed by renal masses. Bowel loops weee not dilated. Aorta and its major branches are patent. The retroverted uterus and bilateral adnexa appeared unremarkable. Urinary bladder was underdistended.

Basal lungs shows bilateral variable sized bilateral pulmonary cysts, likely representing lymphangioleiomyomatosis keeping new patient known tuberous sclerosis history. There was no evidence of pleural effusion. Congestive changes in basal lungs. No pleural or pericardiac effusion.

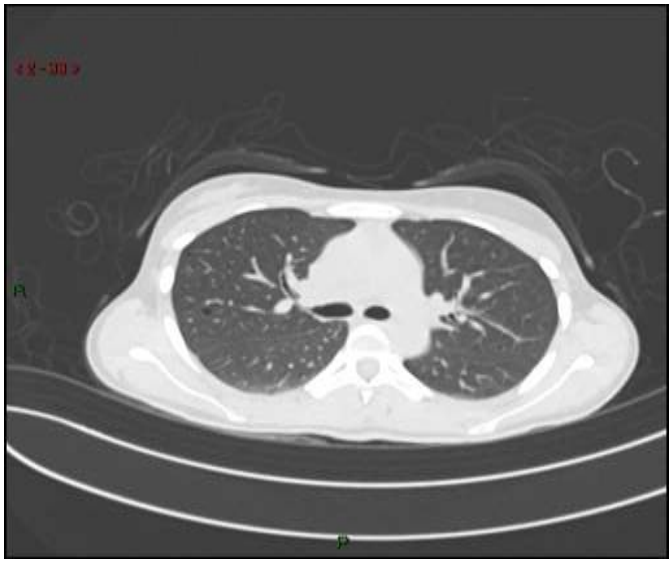

Figure 3: LAM in bilateral lungs.

Few sclerotic foci are noted in posterior elements of L1, T9 and bilateral iliac bones, no osseous destruction or soft tissue component is seen within. The aforementioned bilateral giant renal angiomyolipomas, multiple variable sized bilateral pulmonary cyst and tiny sclerotic osseous foci, all these fall under spectrum of tuberous sclerosis complex.

Considering the large AML and beading of intrarenal arteries, patient was referred to Interventional Radiology department as these have tendency to bleed. No focal target amenable to embolization was seen as there was diffuse involvement of both kidneys and thus case was referred to urologist as she was advised to have bilateral nephrectomy and renal transplantation.

Patient lost to follow up.

Discussion

Tuberous Sclerosis complex (TSC) is a rare, gene-linked multisystem disorder consisting of an autosomal dominant pattern of inheritance. It is manifested as a result of an inactivating mutation in TSC 1 (hamartin) or TSC 2 (tuberin) gene, which, later on, gives rise to tumors of the brain, skin, kidneys, lungs, and retina that may be benign or malignant.[8]

In the general population, the prevalence of TSC is 1 per 600 to 10,000 live births.[9] Essentially, renal disorders are the leading cause of morbidity and mortality in adults suffering from TSC.[10] As it begins during childhood, 55-80% of the patients with TSC acquire a renal angiomyolipoma (AML), with a size-dependent risk of rupture and mortality.[11,12] A study showed that renal lesions were found in 57.5% of the patients, of which 85.4% were diagnosed with an AML.[13]

Renal angiomyolipoma, otherwise known as renal hamartoma, is a rare, benign neoplasm that contains thick-walled blood vessels, smooth muscle, and mature adipose tissue in different proportions. It is one of the many forms in which TSC may manifest.[14,15] The first case of renal AML was reported in 1951, and is more prevalent in females than in males.[12,15] The largest documented giant renal AML was reported by R. Taneja and colleagues in 2013, the size of which was 39 ´ 25 ´ 9 cm.[16]

It appears as an echogenic mass on ultrasound, while the gold-standard imaging modality to investigate and evaluate an angiomyolipoma is computerized tomography (CT) scan, on which it appears to have fat-like density.[17] Once removed, the mass should also undergo histological and immunohistochemical analyses. The annual growth of a renal AML is up to 4 cm in its maximum measurement, but when they grow above 10 cm, they are called 'giant' renal AMLs, which are rarely reported in studies.[14] Signs that indicate the presence of a large AML include abdominal pain, hypertension, a palpable mass, and macroscopic hematuria. When small, it is asymptomatic in most patients.[18]

In a study conducted in 2005, fine-needle aspiration cytology (FNAC) was performed prior to the procedure in 6 cases, 5 of which were misdiagnosed as renal cell carcinoma (RCC).[19]

Angiography showed that TSC-linked AMLs have a vigorous, intricate, and highly invasive blood supply.[20] An expanding AML leads to the formation of hemorrhagic aneurysms, which leads to higher chances of compression symptoms and bleeding from aneurysm rupture.[21,22] A significant risk of rupture requires clinical management of the TSC-linked AML, which may include minimally invasive therapies like transcatheter arterial embolization (TAE), or partial nephrectomy. There's a chance for the AML to render the kidney non-functional, which might even require a total nephrectomy. The rupture of a renal AML may result in severe pain, shock, rapidly advancing anemia, and a fall in blood pressure. These symptoms call for an emergency TAE procedure. Everolimus (an mTOR inhibitor) can also be used to treat TSC-linked AML in high-risk

patients with little to no symptoms, as the side effects of the drug are bearable.[22]

AMLs with complicated blood supply would necessitate embolization of higher complexity that would consequently infarct more of the normal parenchyma of the kidney as collateral damage, which is why the renal function of the patient should be evaluated and put into consideration before as well as throughout the embolization procedure. Large AMLs may be associated with significant morbidities, ranging from flank pain, to renal insufficiency, and even renal failure, because large and they have tendecy to bleed.[23]

In conclusion, radiological analysis via CT scan or ultrasound can help in the visualization of giant renal AMLs, while immunohistochemistry can seal the diagnosis. As our patient has giant bilateral renal AMLs, both with multiple small aneurysms on the intrarenal arteries, her prognosis is worse than of those with unilateral ones.

Conclusion

Large AMLs larger than 20 cm have seldom ever been reported in Pakistan before. These when seen require urgent attention as they have tendency to bleed. Given the significant risk of bleeding associated with this tumor, the preferred course of therapy is a partial or complete nephrectomy in order to minimize bleeding and relieve the symptoms of compression brought on by the large mass. Conservative treatment is not recommended.

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