The Importance Of Renal Angiomyolipoma In The Differential Diagnosis Of The Tuberous Sclerosis

TÜBEROSKLEROZİS KOMPLEKS AYIRICI TANISINDA RENAL ANJİOMYOLİPOMUN ÖNEMİ

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SUMMARY

Renal angiomyolipoma is a benign tumour containing adipose tissue, muscular tissue and vascular structures in different ratios. It consists of 3% of the kidney masses and is rarely seen in childhood. Renal angiomyolipoma is usually asymptomatic and detected incidentally. However, it may be a part of tuberous sclerosis complex disease and is seen in 50-75% of these patients. Thus, tuberous sclerosis complex disease should be excluded in cases with renal angiomyolipoma. We present two cases with renal angiomyolipoma detected incidentally upon evaluation for obesity and nonspecific abdominal pain. Although evaluation for tuberous sclerosis complex disease were negative at the moment, they are being followed up with respect to the development of other criteria for tuberous sclerosis complex disease.

Key words: Renal angiomyolipoma, children, tuberous sclerosis complex disease

ÖZET

Renal anjiomyolipom, yağ dokusu, kas dokusu ve vasküler yapıları değişik oranlarda barındıran, iyi huylu bir tümördür. Çocukluk çağında seyrek görülür ve renal kitlelerin %3'üdür. Renal anjiomyolipoma genellikle asemptomatiktir ve tesadüfi olarak saptanır. Ancak, Tüberosklerozis kompleks hastalağınının bir parçası da olabilir ve bu hastalarda %50-75 oranında görülür. Bu nedenle, renal anjiomyolipomu bulunan olgularda Tüberosklerozis kompleks ekarte edilmelidir. Obezite ve nonspesifik karın ağrısı nedeniyle yapılan tetkiklerinde tesadüfi olarak renal anjiomyolipom saptanan ve Tüberosklerozis kompleks için yapılan değerlendirmelerinde destekleyici bulgu saptanmayan iki olgu, Tüberosklerozis kompleks kriterlerinin takipte de gelişebilecek olması nedeniyle takip edilmektedir.

Anahtar sözcükler: Renal anjiomyolipom, çocuk, tüberosklerozis kompleks hastalığı

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Renal Angiomyolipoma (RAML) is a rare benign tumour composing 3% of the kidney masses. It consists of adipose tissue, muscular tissue and vascular structures in different ratios. Its incidence is 0.1% in males and 0.2% in females (1). Due to lack of apparent symptoms in a majority of the patients, they are often randomly-detected cases.

The main reason of morbidity in RAML is rarely-seen retroperitoneal or spontaneous bleeding that can be as hematuria (2). One of the long-term renal complications is renal failure that usually develops secondary to obstruction. Whereas 80% of the cases are sporadic, 20% of them develop as a part of Tuberous Sclerosis Complex (TSC) disease. Because RAML occurs in 50-75% of TSC patients, TSC disease should be excluded in cases diagnosed with RAML (3). We present two cases with RAML detected incidentally upon evaluation for obesity and nonspecific abdominal pain.

CASE 1

An 8-year old boy presented with the complaint of abdominal pain for the last one year. Abdominal pain was reported to occur once a week, last 1-2 hours and there was no accompanying symptom. Physical examination including anthropometric measurements was normal. Laboratory evaluation including urinalysis, urine culture, complete blood count, blood biochemistry, stool examination for parasite and occult blood, and acute phase reactants were normal. Abdominal Ultrasonography (US) revealed an anechoic cyst, sized 12 mm in lower pole of the right kidney, and a parapelvic hyperechoic angiomyolipoma sized 10 mm in the same lower pole. Abdominal Magnetic Resonance Imaging (MRI), demonstrated both calyceal cyst and angiomyolipoma with increased T2 signal. The renal cyst was having higher T2 signal and lower T1 signal compared to the angiomyolipoma. The angiomyolipoma was not hyperintense on T1 weighted images. (Figure 1a,b). Physical examination was negative for eye and skin lesions of TSC. Brain MRI and echocardiographic evaluations were also normal. We followed up the patient for 30 months and no extrarenal findings associated with TSC developed during follow up.

CASE 2

A 12-year old girl presented for obesity and acnes on her face. Physical examination was characterized by weight 59 kg (1.36 SDS), height 153 cm (0.28 SDS), weight-for-height 131%, blood pressure 120/66 mmHg, and pervasive acne vulgaris lesions on her face. Laboratory tests including urinalysis, complete blood count, blood biochemistry, preprandial blood glucose, thyroid function tests and acute phase reactants were normal. Abdominal US showed a long hyperechogenic angiomyolipoma sized 8 mm in long axis in the lower pole of right kidney. Abdominal MRI depicted the lesion as hypointense on contrast enhanced fat suppressed series (Figure 2). Physical examination was negative for eye and skin lesions of TSC. Brain MRI and echocardiographic evaluations were also normal. We followed up the patient for 49 months and no extrarenal findings associated with TSC developed during follow up.

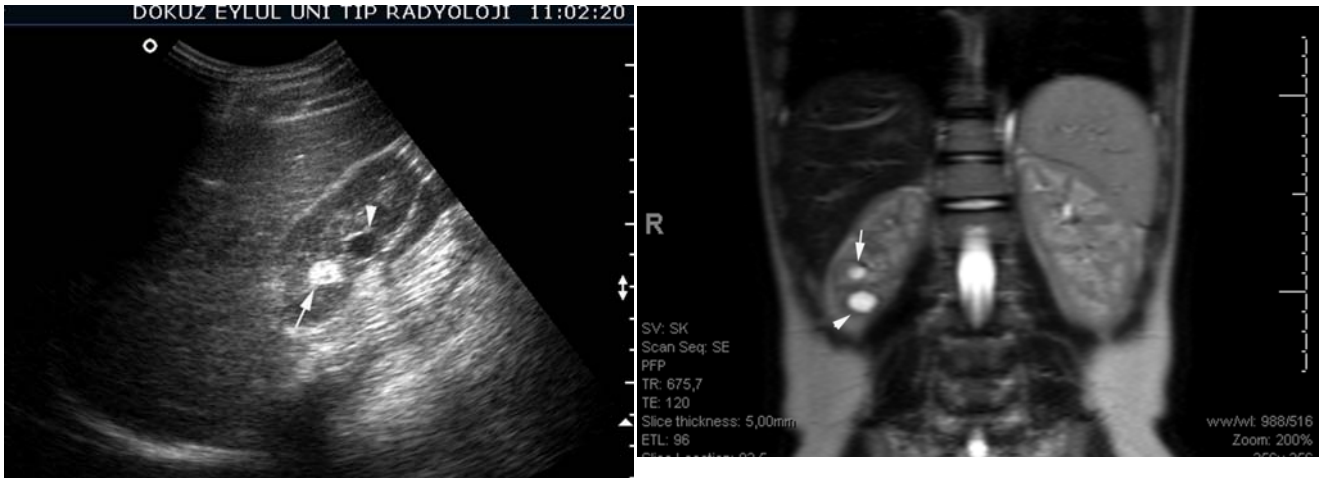

Figure 1. Oblique axial renal US image **(a)** shows classical hyperechogenic angiomyolipoma(arrow) and anechogenic cyst(arrow head). T2 weight coronal renal image **(b)** shows hyperintense focal lesion on the midpart of the right kidney. The renal cyst is seen with higher signal, below the lesion, as well.

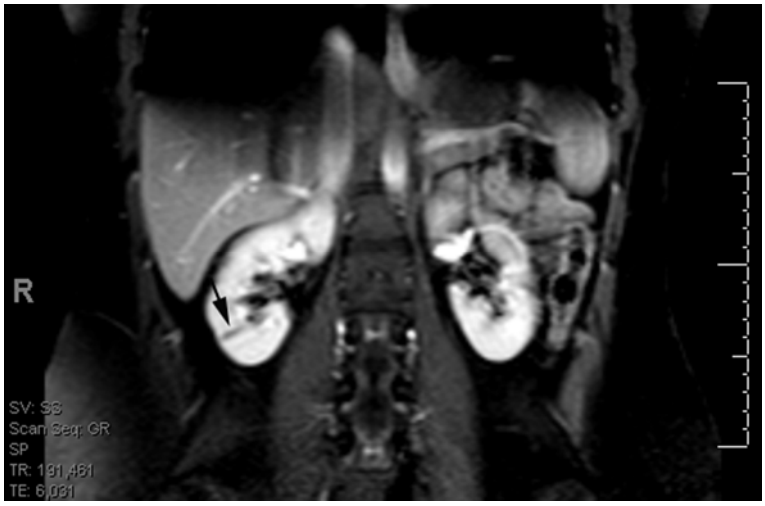

Figure 2. Contrast enhanced T1 weighted fat supressed coronal image shows unenhancing hypointens lesion of the right kidney lower pole (arrow).

DISCUSSION

RAML is a benign tumour consisting of vascular, muscular and adipose tissue. Its frequency is 0.1-0.2 % in general population and is 4-6 times more frequent in girls. It is often detected coincidentaly during evaluation for a non-related symptom. Classical ultrasonographic finding is a renal hyperechoic mass without acoustic shadow (1). The presence of adipose tissue which is characteristic for RAML can be displayed through MRI and computed tomography.

The main morbidity in RAML is spontaneous bleeding that may be retroperitoneal or in the form of hematuria. Although RAMLs over 3.5 cm are known to cause bleeding, there is no established relationship between size of the lesion and bleeding (2). However, symptoms develop in 68-80% of the masses >4 cm, haemorrhage is seen in approximately 50-60% of the symptomatic cases, and hypovolemic shock occurs in one third of the patients with haemorrhage (3).

Renal angiomyolipoma is one of the major diagnostic criteria for TSC and occurs in 8 % of the children below 5 years old with TSC. TSC is an autosomal dominant genetic disorder due to mutations in TSC-1 and TSC-2 genes, characterized by hamartomatous lesions in skin, brain, kidneys, eyes and heart. The prevalence of the disease is 1/6.000-10.000 in general population (4). Presence of facial angiofibromas, non-traumatic ungual/periungual fibromas, hypomelanotic macules, Shagreen patches, retinal hamartomas, cortical tuber and/or subependimal nodules, cardiac rhabdomyomas, lymphangiomyomatosis or renal angiomyolipomas are considered to be major criteria for the diagnosis of TSC (5,6). Among the children over 10 years of age with TSC, RAML could be detected in 50-75%, renal cyst in 17-35% and renal cell carcinoma in 1-2% (1,7,8). Co-occurrence of renal cyst and RAML as in our case 1 was reported in 8 of 207 cases (4%) evaluated due to TSC (3).

Subependimal nodules are seen in 82.9%, 96.6% and 100% of the children with TSC younger than two years, 5-9 years, and 9-14 years of age, respectively (9). Most frequent cardiac finding is rhabdomyoma which is present in 50-80% of cases (10). Ophthalmic findings such as retinal tumours, pigmentary changes in iris and colobomas can be seen in patients with TSC. Physical examination was negative for eye and skin lesions of TSC in our patients. Their brain MRIs and echocardiographic evaluations were also normal. Review of the current literature disclosed no case with initial finding of renal angiomyolipoma who was diagnosed as TSC in follow up. However, cases with isolated RAML should also be followed up for extrarenal findings of TSC since accompanying findings could de-

velop in elder age groups (6,9). We followed up case 1 and case 2 for 30 and 49 months, respectively. Neither of them developed extrarenal findings associated with TSC during follow up.

Tuberous sclerosis complex patients should be followed up periodically as they are at risk for RAML associated renal parenchymal hemorrhage and chronic kidney disease (4). Follow up and treatment of these patients depend on the presence of symptoms and size of the RAML. Selective arterial embolization or partial nephrectomy is treatment options for symptomatic tumours bigger than 4 cm. Asymptomatic tumours in this size should be monitored thorough imaging by USG every six months. Symptomatic tumours <4 cm should also be monitored since symptoms may regress; however, if there is no regression of the symptom, embolization or partial nephrectomy should be tried. Asymptomatic tumours <4 cm should be monitored every 1 to 3 years regularly (11). Since RAML sizes were <4 cm and no symptom associated with RAML was present in our cases, we followed up case 1 and 2 by serial USG for every six months that showed no change in the size of the lesion.

In conclusion, children with RAML should be followed up both for the complications associated with the renal tumour and also for the development of extrarenal findings of probable TSC.

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