Tuberous Sclerosis with Angiomyolipoma and Renal Cell Carcinoma

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Abstract

Angiomyolipomas and renal cysts are the common renal lesions associated with Tuberous Sclerosis. Reported incidence of Renal Cell Carcinoma (RCC) in Tuberous Sclerosis is only 2%. We report a case of Tuberous Sclerosis with bilateral angiomyolipomas and left Renal Cell Carcinoma.

A 30-yr-old female presented with history of pain and mass in the left lumbar region of 3 months duration. She had undergone laparotomy and left partial nephrectomy at a local hospital. Histopathology showed sarcomatoid variant of RCC. On examination, she had features of Tuberous Sclerosis such as Adenoma Sebaceum over the face, Subungal Fibroma and white leaf shaped macules over the thigh.

Keywords: Tuberous Sclerosis, Angiomyolipomas, Renal Cell Carcinoma, Adenoma Sebaceum

Introduction

The renal lesion usually associated with Tuberous Sclerosis is Angiomyolipoma. Renal cyst is the next most commonly found renal lesion.1 The reported incidence of Renal Cell Carcinoma in Tuberous Sclerosis is 2-4%.2 We report a case of Tuberous Sclerosis with bilateral renal Angiomyolipomas and left Renal Cell Carcinoma.

The Case

A 30-year-old female presented with the history of pain and mass in the left lumbar region of 3 months duration. She had undergone exploratory laparotomy one year back for a mass in the left iliac and lumbar regions with a provisional diagnosis of ovarian cyst at a local hospital. Discharge summary revealed histopathology report showing sarcomatoid variant of Renal Cell Carcinoma. She gave history of epileptic attacks during childhood.

On examination, the patient had all the classical features of Tuberous Sclerosis: Adenoma Sebaceum on the face, Subungal Fibroma over right index finger (Koenen tumor), Semi lunar hypopigmented patches over back and Ash-Leaf macules over the left thigh (Figures 1-3).

Per-abdominal examination showed a right paramedian scar and bilateral large ballotable flank masses. Higher mental functions were normal.

Investigations

Urine examination showed 1 to 2 RBC’s/hpf. Haemoglobin was down to 9.0g/dl and ESR was 40 mm in the first hour. Chest X-ray was normal.

Abdominopelvic Ultrasound showed that Left kidney was enlarged with a large (7x6cm) hypoechoic lesion in the upper pole. Right Kidney was also enlarged and outline deformed.

Figure 1. Adenoma Sebaceum over the face

Figure 2. Subungal Fibroma over right index finger (Koenen Tumor)
with multiple hyper echoic regions all over the parenchyma. C T Abdomen showed a large (9x6x7cm) necrotic mass in left lumbar region infiltrating and encasing splenic flexure and upper descending colon. Multiple enlarged lymph nodes were seen in left renal fossa, para-aortic region and retro-crusal region. Multiple fat density lesions were seen in both kidneys suggestive of angiomyolipomas (Figures 5 & 6).

C T Brain showed calcified subependymal hamartomas in the lateral ventricular surface and right temporal horn (Figures 7 & 8). Echocardiography showed mild Left Ventricular Hypertrophy, and no evidence of Rhabdomyoma (Figure 4).

Histopathology showed cystic and necrotic areas with sarcomatoid elements made up of spindle cells admixed with epithelial component. Cells had irregular nuclei with nucleoli. Also observed were clear cell areas admixed with stromal components.

Treatment

In view of the extensive disease, when the patient and their attendants were explained the risks of attempting nephron sparing surgery and probable need for renal replacement therapy later, they were not willing for any surgical intervention. So, palliative chemotherapy with vinblastine was started.

Discussion

Tuberous Sclerosis is an inherited, autosomal dominant neurocutaneous disorder characterized by seizures, mental retardation, cutaneous lesions and visceral hamartomas described by Bourneville in 1880. In incidence is 10.6 cases /1,00,000 persons.
Lesions associated with tuberous sclerosis are as follows:

- **Brain**: Cortical tuber
- **Retina**: Phakomas
- **Skin**: Angiofibroma
- **Heart**: Rhabdomyoma
- **Bone**: Sclerotic lesions
- **Lung**: Lymphangioma
- **Kidney**: Angiomyolipoma and renal cysts

Renal angiomyolipomas occur in 50 – 80% of patients and are usually small, multiple and bilateral. Large angiomyolipomas are more likely to bleed and prophylactic embolisation or surgical excision is recommended if size is > 4 cm. Renal cysts develop in about 20% of patients. The cystic disease can lead to renal failure, probably because of compression of the parenchyma by the expanding cysts, which is uncommon before 4th decade.

Biopsy of renal cysts in Tuberous Sclerosis is pathognomonic, showing eosinophilic epithelial hyperplasia lining the cysts. The incidence of Renal Cell Carcinoma in Tuberous Sclerosis complex has been reported to be 2-4%, which is higher than that of the general population. These occur at a younger age (mean 28 yrs), 80% occur in women and 43% are bilateral. 50% of lesions show high-grade sarcomatoid features. Renal cell carcinoma evolves from the cystic epithelium.

**Conclusion**

Tuberous sclerosis is a lifelong disease and patients are at risk of developing malignancies and hence patients need to be regularly followed up. Other members of the family also need to be screened for the disease.

**End Note**

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**Conflict of Interest** – None Declared

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