Abstract:
Neurocutaneous syndromes are rare disorders that result in lesions in both the central nervous system and skin. Tuberous sclerosis is an important neurocutaneous syndrome with urological implications ranging from benign cysts to renal cell carcinomas. Herein we report a 40 year old lady who presented with bilateral large renal masses with classical features of Tuberous Sclerosis.

Keyword: Tuberous sclerosis, Angiomyolipoma

Introduction:
Neurocutaneous syndromes are disorders of central nervous system that additionally result in lesions on the skin and the retina. Tuberous sclerosis is the second most common neurocutaneous syndrome that is inherited in an autosomal dominant fashion with a live birth prevalence of 10-16 cases per 1,00,000. Renal manifestations include renal cysts, angiomyolipomas and renal cell carcinoma. Approximately 50% of patients with TSC develop angiomyolipomas.

Case report:
40 year old lady presented with H/O bilateral loin pain since 1 month associated with generalised weakness. Loin pain was dull aching type with no radiation of pain noted. No history of hematuria/ weight loss, loss of appetite. There were no voiding symptoms. No history of past surgeries. Patient has regular menstrual cycles and one living child 18 years of age.

On Examination
Patient was moderately built and nourished. There were multiple hyper pigmented papules in malar region of face (Figure 1). Further examination revealed shagreen patch in the back (Figure 2), and multiple subungual fibromas (Figure 3). Examination of the abdomen revealed fullness in both lumbar regions on inspection. On palpation a 8X10 cm soft to firm mass with irregular surface was felt in both lumbar regions moving with respiration. The mass was bimanually palpable. On auscultation there was no bruit heard.
Investigations:
Hb – 11 gms TC – 4500 cells/cmm RFT – Within normal limits Urine Albumin/sugar – nil Deposits 1-2 pus cells and 1-2 RBC’s Urine C/ S - No Growth X-ray KUB – NAD USG KUB: mixed heterogenous mass seen in both the kidneys with areas of hyper echogenecity. No evidence of thrombus seen in the renal veins or IVC. CECT Abdomen: Bilateral mixed density lesions seen involving both the kidneys with minimal enhancement on contrast study (Figure 4).

Figure 4: Mass lesions in both kidneys
Coagulation profile was normal and patient underwent USG guided trucut biopsy of the renal mass. Patient had mild hematuria for two days following the procedure that was managed conservatively.

Histopathology report:
Microscopic description: Section showed linear fragments of renal tissue with glomeruli. Linear bits of tumor tissue showing spindle cells in fascicles interspersed with blood vessels that show hyalinisation of the walls were also seen. Few foam cells seen within the lesion. Features suggestive of Bilateral renal Angiomyolipoma (Figure 5).

Management plan:
Patient is planned for angioembolisation of the feeder vessels of the renal mass and renal sparing surgery at a later date.
Discussion:
Tuberous sclerosis is a rare autosomal dominant disease characterised by the triad of adenoma sebaceum, mental retardation and seizures. Diagnosis of Tuberous Sclerosis made on the basis of major and minor criteria. Adenoma sebaceum, Angiomyolipoma and Shagreen patch seen in this patient are part of major criteria. AML greater than 4 cms and symptomatic have to be surgically excised. Nephron sparing procedures have to be done in view of bilateral and multifocal tumors. This patient is being planned for angioembolisation of the feeder vessel and preservation of viable renal tissue if possible. Patient has been counselled regarding need for bilateral nephrectomy and renal replacement therapy and transplantation in the future.

References: