20-year-old female with past medical history of repeated seizures, presented with haematuria and right loin pain. Clinical examination revealed at least three classical hypopigmented macules on lower abdomen (Ash leaf lesion) with Shagreen patches and angiofibromas over lower back. Fig A&B: FLAIR and T2 images of Brain showed cortical/subcortical tubers and subependymal hamartomas. Fig. C: CT chest revealed numerous pulmonary cystic foci compatible with lymphanioleiomyomatosis. Fig D: Multiple sclerotic bone lesions in CT scan of thoracic spine.

Fig E-G: CT and MR scan (T1 and Fat Saturated images) of the abdomen show angiomyolipomas in the liver and left kidney. These lesions exhibit fat density on CT and follow fat signal on T1 and Fat saturated images.

A confident diagnosis of Tuberous sclerosis was made as the patient’s clinical features and radiological features fulfilled 5 major and 2 minor criteria for TSC.

Tuberous sclerosis (TS) also known as Bourneville’s disease is the second most common neurocutaneous syndrome after neurofibromatosis with autosomal dominant inheritance. A confident diagnosis of TS can be made using Tuberous sclerosis complex (TSC) diagnostic criteria which includes several clinical and radiological findings. At least 2 major or 1 major with 2 or more minor features are required for a definitive diagnosis of TS. A vast majority of patients (96%) have skin manifestations with variations based on age. Within the brain, there are three typical types of nodular lesions which include subcortical heterotopic nodules, cortical tubers and subependymal giant cell astrocytomas. In skeleton, if more than four sclerotic bone lesions are detected on CT scan of any main body region, the differential-diagnosis of TSC has to be considered.

References