Tuberous Sclerosis with Bilateral Angiomyolipoma

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A 26 years old female presented with bilateral dull aching flank pain and hematuria for last two months. There was no h/o headache, vomiting, seizure, trauma, abnormal behaviour, cognitive decline or bladder-bowel involvement. Her father and brother also had similar skin lesions on forehead and face. General physical examination revealed multiple adenoma sebaceum and a fore head plaque on her face (Figure 1A).

Her neurological examination (MMSE -26/30) and biochemical investigations were unremarkable except the urine which showed RBC cells 10-15 /cubic mm without microalbuminuria and sugar. Contrast enhanced CT-abdomen revealed huge heterogeneously enhancing mass lesions with fat density in both kidneys, right and left kidney measuring 22.3 x 22 mm and 67 x 34.8 mm respectively (Figure 1B). Magnetic resonance imaging (MRI) brain with contrast showed subependymal nodules, cortical and subcortical tubers, white matter hyperintensities and giant cell astrocytoma (SEGA) (Figure 2). Diagnosis of tuberous sclerosis was made on the basis of recommendations of the 2012 International Tuberous Sclerosis Complex Consensus.¹ Our patient had atypical presentation despite all radiological findings of tuberous sclerosis, large forehead plaque and the positive family history of skin lesions without any neurological sign and symptoms.

References