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Tuberous sclerosis: Renal, skin and brain involvement

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Description

21-year-old male presented to our pulmonology clinic with complaints of abdominal discomfort for 2 weeks. He had history of seizures since childhood well-controlled with anti-epileptic medications. Patient had lesions on face and back on physical examination (Figure 1A & 1B). There were no other positive findings on examination and his family history was also insignificant for any hereditary illness.

Patient underwent Contrast Enhanced Computed Tomography (CECT) of abdomen along with other laboratory investigations. Axial cuts of CECT abdomen revealed right renal mass with areas of focal fat density and multiple enhancing vessels coursing through it suggestive of Angiomyolipoma (AML) (Figure 1C). Brain Magnetic Resonance Imaging (MRI) done for evaluation of seizures showed T1 hyperintense subependymal lesions in bilateral lateral ventricles suggestive of ependymomas (Figure 1D). Based on classical clinical features diagnosis of Tuberous Sclerosis Complex (TSC) was made. Evaluation for involvement of other organs (heart, lungs and eyes) were negative. In view of solitary renal AML of size <6 cm surgery was deferred and surveillance imaging was offered. Interval surveillance imaging was also suggested for monitoring brain lesion. Laser therapy and mammalian Target of Rapamycin (mTOR) inhibitor everolimus was denied by patient for skin and renal AML.

TSC is a rare genodermatosis with birth incidence ranging from 1:6000 to 1:10000. It is characterized by development of benign tumors (Tubers) throughout body. Major morbidity and mortality in this condition is due to renal and neurological involvement.

Renal AML and ependymomas occur in around 80% cases of TSC [1]. mTOR pathway is central to formation of tumors in TSC.

mTOR inhibitors like everolimus are approved for management of renal AML and brain lesions in TSC especially in those cases where surgery is not feasible [2]. Angioembolization of vessels for renal AML is another option where chances of bleed due to vascularity is higher. Laser therapy is offered for management of skin lesions. Early diagnosis and treatment directed to renal and neurological manifestations significantly improve outcomes.

In this article a rare condition of tuberous sclerosis is described [3]. Classical manifestations along with treatment options available for renal manifestations are highlighted.

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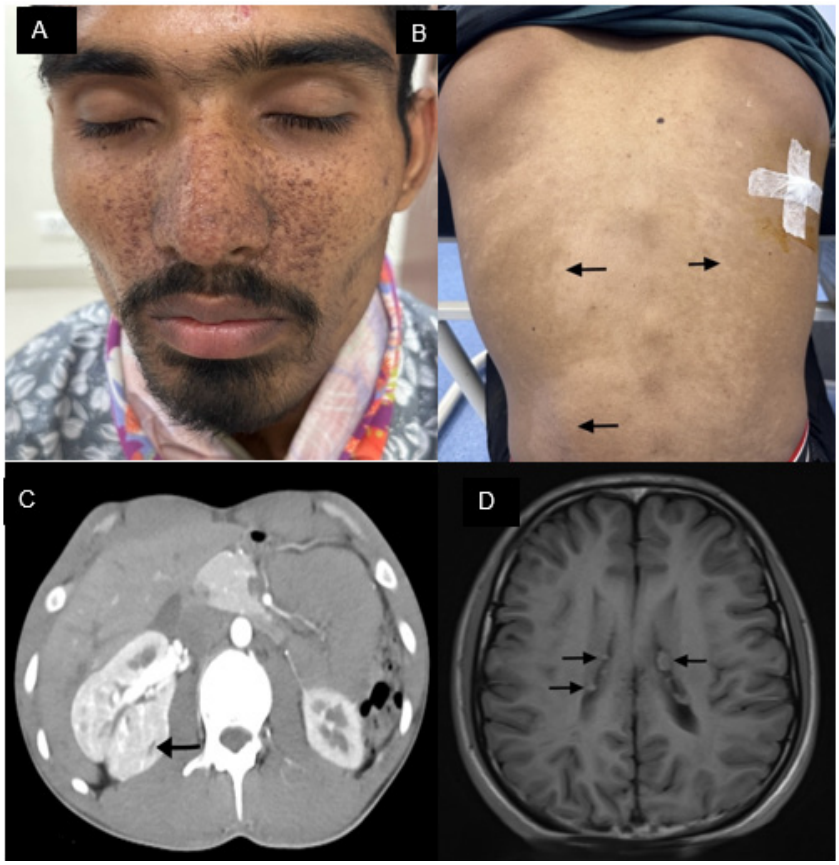

Figure 1: A) Erythematous papules over face of 21-year-old patient consistent with angiofibroma. **B)** Hypomelanotic macules over upper back and connective tissue nevi suggestive of shagreen patch over lower back of our patient. **C)** CECT abdomen axial cut demonstrates mass in right kidney, fat within lesion is demonstrated as low attenuation area (arrow) suggestive of angiomyolipoma. **D)** Brain MRI axial T1 sequence showing subependymal lesions in lateral ventricles (arrow) consistent with ependymoma.