Differing Presentations of Familial Tuberous Sclerosis

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A 18-year-old girl presented with history of diminution of vision for the past 3 months, headache on and off since 2 months, and vomiting since 3 days. No history of fever, seizures or trauma. She has had mental retardation since childhood. She is the third daughter (out of four - others normal) born of non-consanginous marriage. On examination, the patient was conscious, oriented, and had gingival hypertrophy and neurocutaneous markers (adenoma sebaceum and shagreen patches). Her CNS examination was otherwise normal. Clinically, a diagnosis of Tuberous Sclerosis was made¹. The mother of the patient, 40 years old, also had adenoma sebaceum on her face but was asymptomatic. Investigations revealed presence of Subependymal Giant Cell Astrocytoma, Subependymal nodules/hamartomas and multiple sclerotic bone lesions, in both the patient and the mother. The patient had obstructive hydrocephalus² and Optic atrophy, and a mass in the heart suggestive of cardiac rhabomyoma (Figure 1). The mother, though completely asymptomatic had an Angiomyolipoma in the left kidney, Bilateral simple renal cysts, Hepatic angiomyolipoma and lymphangioleiomyomatosis in lungs (Figure 2). The patient underwent symptomatic treatment and was then referred to neurosurgery for Ventriculoperitoneal shunt surgery. We report a mother and daughter with differing presentations in a case of Familial Tuberous sclerosis with no history of seizures.

References
