

Photoclinic



Figure 1 (A and B). Facial lesions and a hypopigmented spot on the patient's back.

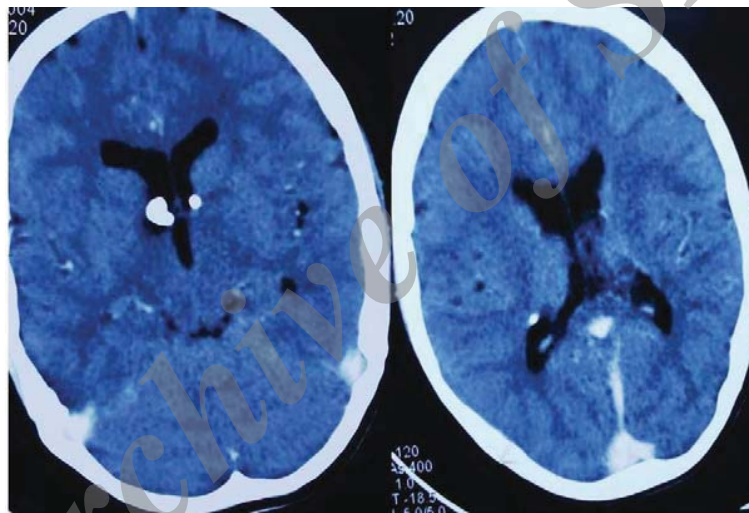


Figure 2. CT of the head showing subependymal nodules.

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A 16-year-old male presented in an emergency department with recurrent episodes of generalized tonic clonic seizures. The convulsions were controlled with intravenous diazepam (10 mg) and the patient was administered a loading dose of 800 mg of phenytoin over half an hour. Work up for metabolic causes that included random blood sugar, hemogram, renal and liver function tests and serum electrolytes (sodium, calcium and magnesium) were normal. The patient was a known epileptic and had been on oral

phenytoin for the past five years. Since he had been seizure-free for the past two years he had recently discontinued the medication by himself. The patient had never been to school. According to his mother, he was less bright than his other siblings. The patient had peculiar facial lesions (Figure 1A) and some hypopigmented macules on his back (Figure 1B). Contrast enhanced computed tomography of brain was performed (Figure 2). No other family members had similar lesions or any history of epilepsy. What is your diagnosis? See the next page for diagnosis.

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What is your diagnosis?
See the next page for diagnosis.

Photoclinic Diagnosis:

Tuberous sclerosis

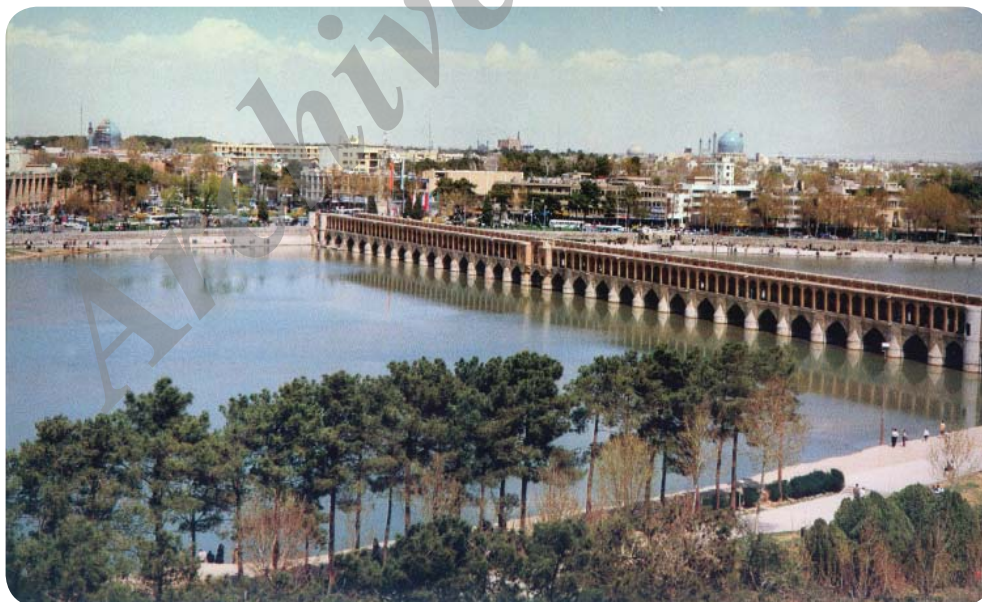
Tuberous sclerosis is an autosomal dominant inherited disease which can have variable presentations. However, in the majority of cases it results from new mutations. Two genes believed to have a role in causation have been mapped to chromosomes 9q34 (TSC1, Tuberin) and 16p13 (TSC2, Hamartin).¹ It is also recognized by the terms epiloia (after the clinical triad of epilepsy, low intelligence, and adenoma sebaceum) and Bourneville disease. While cases are frequently diagnosed in the first decade of life, they may also present late. Adenoma sebaceum (angiofibromas) are the characteristic cutaneous lesions (Figure 1A) that usually involve the nasolabial folds, however they may involve other areas such as the cheeks, forehead, and scalp. They manifest early and increase until adolescence. Other cutaneous manifestations include periungual fibromas (Koenen tumors), Shagreen patches or ash-leaf macules. Shagreen patches are soft and fleshy plaques are usually located in the lumbosacral area.² Ash leaf-shaped macules are ovoid, hypopigmented macular lesions found on the trunks or limbs (Figure 1B). They can be appeared even at birth. Neurologic features include epilepsy and mental retardation which result from the occurrence of cortical tubers. Tubers can occur at other sites such as the cerebellum and spinal cord, and the manifestations depend on their location, size and growth. Subependymal nodules are also seen. These are located usually in the wall of lateral ventricles and may have calcifications (Figure 2). An increase in size suggests degeneration into a subependymal giant cell astrocytoma (SEGA). Other findings may include car-

diac rhabdomyomas, aortic aneurysms, renal angiomyolipomas, and pulmonary lymphangiomatosis.²

The goals of therapy are to provide the best quality of life. Vigabatrin can be used for infantile spasms. Carbamazepine, oxcarbazepine, and phenytoin can be utilized to treat partial seizures in adolescents and adults. SEGA can be treated with surgical resection. Everolimus and rapamycin may be used in non-resectable SEGA and act by inhibiting the growth of TSC-deficient cells.³ They may also be of benefit in renal angiomyolipomas.⁴ Clinicians should be aware of the possibility of tuberous sclerosis in patients who present with seizures. Usually the cutaneous manifestations will determine the diagnosis to the astute clinician.

References

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Sio-Se-Pol [Allah-Verdy-Khan Bridge; Safavids Era (1501–1722)], Isfahan, Iran. (Source: 'A Glance at Esfahan, City of Art' by Parviz Dabiri MD, Mehr Afrouz Publication, Isfahan, Iran, 2005; p: 162)