

PHOTOCLINIC



Figure 1

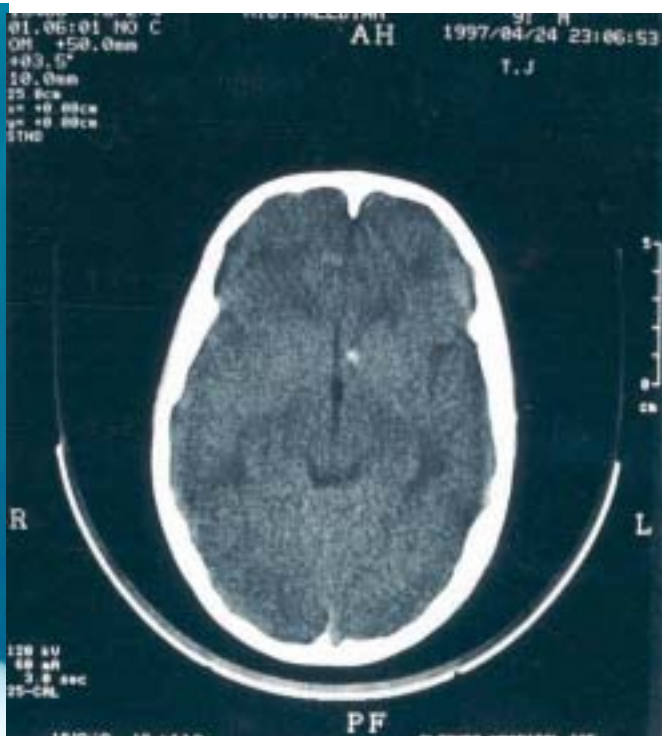


Figure 2.

A 14-year-old boy was admitted to the outpatient department with seizure. On admission, there were red-to-pink nodules over the nasolabial folds, cheeks and chin (Figure 1). There was no evidence of mental retardation, and electroencephalography showed epileptiform discharges. Plain radiography and computerized tomography scanning (Figure 2) disclosed intracerebral calcification.

Your Diagnosis?
See page 268 for diagnosis

Photoclinic Diagnosis: Tuberous Sclerosis (Epiloia-Bourneville's Disease)

Tuberous sclerosis complex (TSC) is an autosomal dominant disease involving predominantly the skin and nervous system. The prevalence of the disease is estimated to be 1 to 15 per 100,000 population.¹

Seizure, mental retardation and skin lesions are among the most common symptoms of the disease; infantile spasm is particularly frequent in young adults.^{2,3}

The cutaneous lesions in TSC include:

- 1) Hypomelanotic macules of 1 to 2 cm in diameter with an ash-leaf spot scattered randomly on the trunk or extremities. These lesions may be present at birth or may develop in early childhood.⁴
- 2) Angiofibromatous lesions, most often appear over the cheeks and forehead, are rarely present at birth, and are usually first observed between the ages of 1 and 5 years.^{4,5}
- 3) Ungual, or periungual fibromas most often appear at puberty and continue to develop with age; a common site of fibromatous involvement is the nail bed.^{5,6}
- 4) Shagreen patches, usually present over the lumbosacral or gluteal region, rarely present before puberty. These lesions are found in only 20 – 30% of patients.⁴

In the brain, three types of lesion occur: cortical tubers, subependymal nodules and disorders of myelination. The most characteristic gross abnormalities are the presence of tubers. Tubers can be located in the convolutions of any part of the cerebral hemispheres.^{6,7}

In approximately one-half of patients, calcium is deposited within the tubers to an extent as to be visible on plain radiography of the skull or on CT scanning.³

Subependymal nodules are found in the

ventricular walls, particularly in the region of the foramen Monro or the aqueduct of Sylvius, and may result in hydrocephalus. Calcification of these nodules is common and increases with age.⁴

There is no specific treatment. The epilepsy is treated with anticonvulsant drugs. The infantile spasm responds well to vigabatrin, but there are no other known specific responses to particular antiepileptic drugs. Cerebrospinal fluid shunting procedure from the ventricles is the mainstay of therapy for hydrocephalus.⁸

References

- 1 Menkes JH, Sarnat HB. *Child Neurology*. 6th ed. Philadelphia: Lippincott William's & Wilkins; 2000: 865 – 72.
- 2 Jozwiak S, Goodman M, Lamm SH. Poor mental development in patients with tuberous sclerosis complex: clinical risk factors. *Arch Neurol*. 1998; **55**: 379 – 84.
- 3 Riikonon R, Simell O. Tuberous sclerosis and infantile spasms. *Dev Med Child Neurol*. 1990; **32**: 203 – 9.
- 4 Merritt HH, Rowland LP. *Merritt's Neurology*. 10th ed. Philadelphia: Lippincott Williams & Wilkins; 2000: 1069 – 72.
- 5 Miller SV, Roach ES. Neurocutaneous syndrome. In: Bradley WG, ed. *Neurology in Clinical Practice*. 3rd ed. Boston: Butterworth-Heinemann; 2000: 1666 – 71.
- 6 Shepherd CW, House WO, Gomez MR. MR finding in tuberous sclerosis complex and correlation with seizure development and mental impairment. *Am J Neuroradiol*. 1995; **16**: 149 – 55.
- 7 Shepherd CW, Gomes MR. Mortality in Mayo Clinic tuberous sclerosis complex study. *Ann New York Acad Sci*. 1991; **615**: 375 – 7.
- 8 Shorvon DS. *Handbook of Epilepsy Treatment*. Malden, MA, USA: Blackwell Science; 2000: 63 – 5.

A. Ghorbani MD, Department of Neurology, Isfahan University of Medical Sciences, Isfahan, Iran.