

## Tuberous Sclerosis

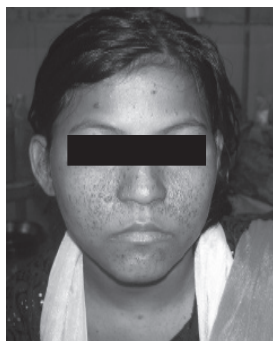
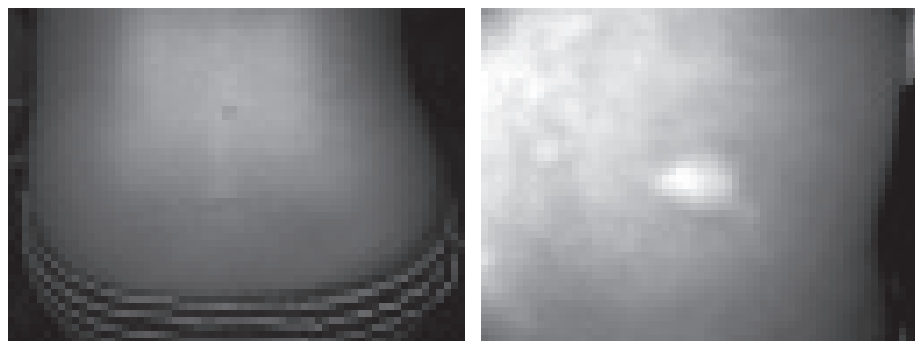
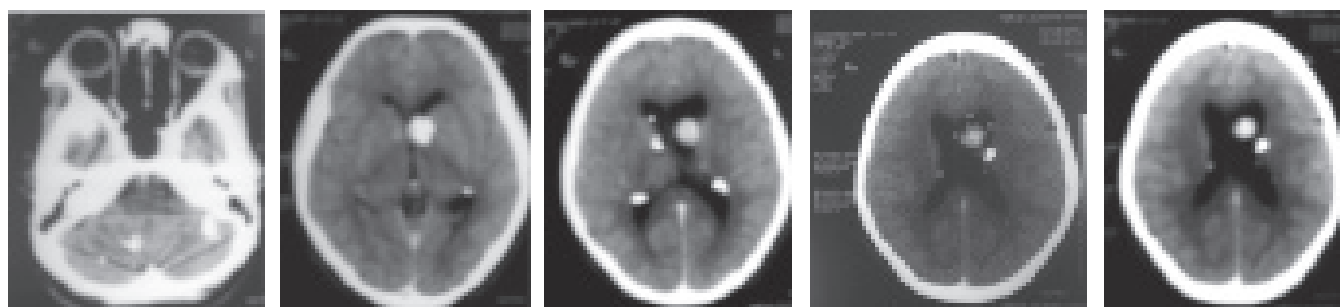


Fig. 1 : Adenoma sebaceum.



Figs. 2 and 3 : Shagreen patches.



Figs. 4 to 8 : CT brain showing subependymal nodules, cerebellar tubers, giant cell astrocytoma and hydrocephalus.



Fig. 9 : Splenomegaly.

A 17 years girl presented with multiple yellowish – red popular lesions over nasolabial folds and cheeks suggestive of adenoma sebaceum and shagreen patches at lumbar region. Her intelligence was normal and she had no history of seizures. She had no headache, vomiting, fever and any focal neurological signs. She had moderate splenomegaly. Blood pictures and malaria parasites were normal. CT brain showed multiple slightly enhancing subependymal nodules and some cortical tubers in cerebellum with calcifications. There was a well-defined enhancing lesion near the foramen of Monro measuring 1.3cm\*1.5cm suggesting giant cell astrocytoma. Hydrocephalus was also present. USG showed moderate splenomegaly without any focal lesion and other findings were normal. Echocardiography was normal. Chest X-ray was normal. EEG was normal.

The classic triad of tuberous sclerosis includes mental retardation, epilepsy and skin lesions. In infancy it is the commonest identifiable cause of salaam attacks. Most patients with mental retardation have epilepsy. Patients with normal intelligence are increasingly being recognized. Other features include depigmented naevus or ash leaf macule, café-au-lait spot, sub and periungual fibromas. Shagreen patch, in reality a plaque of subepidermal fibrosis is seen in 20-30% cases. Many of the clinical features of tuberous sclerosis result from hamartomas; true neoplasm also occur; particularly in the kidney and brain. Ophthalmoscopy may reveal multiple retinal hamartoma. Neuropathological lesions of tuberous sclerosis include subependymal nodules, cortical hamartomas, areas of focal cortical hypoplasia, and heterotopic gray matter. Subependymal nodules are demonstrated on CT where their calcific nature has led them to be termed 'brain stones'. Cerebellar anomalies can be seen in more than one fourth of patients with tuberous sclerosis. Subependymal giant cell astrocytomas (SEGAs), which may develop near the foramen of Monro, may enlarge and cause symptoms of increased intracranial pressure.<sup>1</sup> Hydrocephalus may also be non-specific.

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### REFERENCE

1. Torres OA, Roach ES, Delgado MR, *et al.* Early diagnosis of subependymal giant cell astrocytoma in patients with tuberous sclerosis. *J Child Neurol* 1998;13:173-7.