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Tuberous Sclerosis in Child

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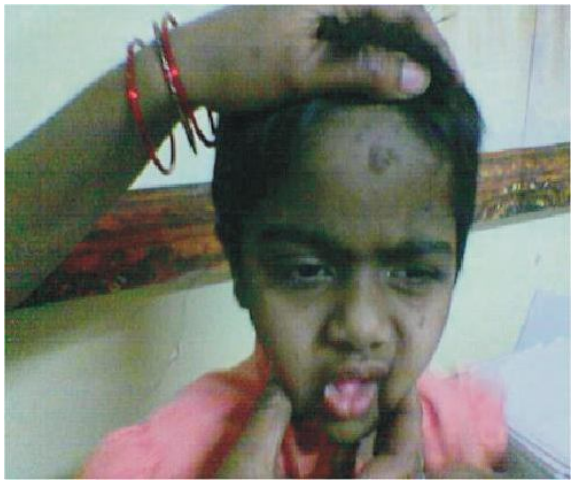
Sir,

A 9 yr, female child presented with history of generalised episode of tonic clonic convulsion since the age of 6month with global developmental delay. Multiple tiny red nodules were present on the forehead (Fig1.), cheeks and nose, suggestive of sebaceous adenoma. Whereas well circumscribed hypopigmented lesion 4-5 in number measuring 3-4cm were noticed over trunk and extremities. CT scan revealed calcified tuber in periventricular areas. The echocardiogram and ultrasonographic examination of abdomen were within normal limits. EEG revealed abnormal electrical discharges. In view of clinical details, CT scan and EEG, the child was diagnosed as a case of Tuberous sclerosis. She was prescribed vigabatrin, sodium valproate, and Multivitamins. The seizures were controlled drastically after starting anticonvulsant therapy.

Fig 1: Presence of Sebaceous Adenoma on the Forehead



Tuberous sclerosis (TS) Epiloia Or Bournervlle's Disease is an autosomal dominant neuro-cutaneous syndrome characterized by abnormalities of both the integument and centre nervous system (CNS) with an estimated frequency of 1/6000.[1-2] The TS gene is located on chromosome 9q34 (TSC) and 16 p 13 (TSC2), but at least half of the cases are sporadic owing to new mutations.[2] TS is an extremely heterogeneous disease with a wide clinical spectrum varying from severe mental retardation and incapacitating seizures to normal intelligence and a lack of seizures, often within the same family.[1] Diagnosis of the tuberous sclerosis should be suspected in all cases of infantile spasm. Skin should be examined in each case for hallmark of the disease if possible, under wood's lamp. The typical skin lesion are hypopigmented 'ash leaf' macules, shagreen patch, subungual and periungual fibroma and sebaceous adenoma which appear at variable ages. Approximately 50% of children with TS have rhabdomyoms of the heart.[3] The disease affects many organ systems other than skin and brain including

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the heart, kidney, eyes, lungs and bone. The diagnosis is established on head CT scan or an MRI in most cases showed calcified tubers in periventricular region and cortical hypodensities.[4] Molecular studies are beneficial in identifying familial cases for a definite prenatal diagnosis. To conclude any child who presents with seizures must be looked for skin lesions.

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