CASE REPORT

Tuberous Sclerosis in a Child

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Abstract

Tuberous sclerosis is one of the important neurocutaneous syndrome chracterized by abnormalities of both the integument and central nervous system. We present here a classical case of tuberous sclerosis. This is a three year female child who had myoclonic seizures, delayed milestones and had hypopigmented skin lesions. MRI brain and EEG were consistent with the diagnosis.

Key Words

Tuberous sclerosis, Neurocutaneous syndrome

Introduction

Tuberous sclerosis (TS) Epiloia Or Bournervlle's Disease is one of the important neuro-cutaneous syndrome characterised by abnormalities of both the integument and centre nervous system (CNS) with an estimated frequency of 1/6000 (1-4). 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). The younger the patients present with symptoms and signs of TS, the greater is the likelihood of mental retardation. The disease affects many organ systems other than skin and brain including the heart, kidney, eyes, lungs and bone (1-5). We present here a child who had classical features of tuberous sclerosis.

Case Report

This was a 3 year old child who presented with seizures for last 2 years. The child was already on multiple anticonvulsants. On detailed history, the seizures were like myoclonic jerks. The child did have delayed mental and motor milestones. There was no history of trauma, fever, ear discharge, vomiting and loose motions etc. Except seizures the child did not have any symptoms pertaining to heart, kidney, eyes and lungs. There was no family history of seizures. On detailed examination, the child was malnourished (PEM grade II as per Indian Academy of Pediatrics) with mild pallor. The child did have hypopigmented skin lesions on face, abdomen and trunk (Fig.1&2). Also a roughened, raised lesion with an orange peel consistency was seen on right side of forehead (Fig.1).

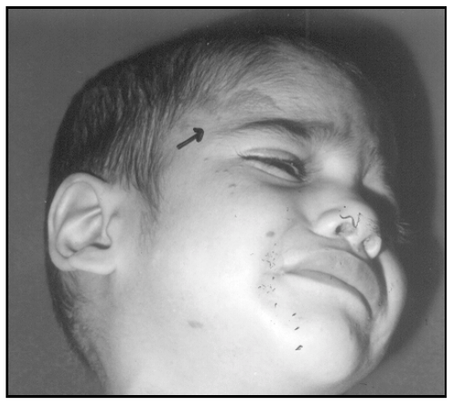

Fig. 1. Roughened, raised lesion on forehead

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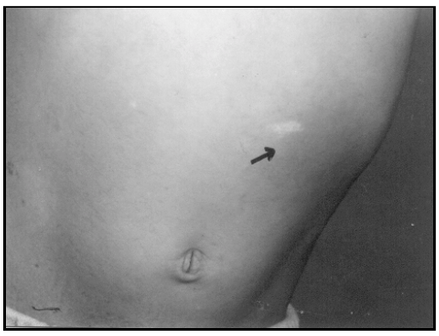

Fig. 2. Hypopigmented skin lesions

It was a home delivery born after consangious marriage. Antenatal and perinatal history was uneventful. Immunization and feeding history was satisfactory. Child used to get myoclonic jerks during examination. Detailed systemic examination and fundus examination did not reveal any abnormality. The child was subjected to investigations. Hemoglobin was 9gm/dl. The peripheral smear depicted normocytic normochromic type of anemia. Blood sugar serum calcium, phosphorous, liver function and renal function tests were all with in normal limits. X-ray skull & chest were also within normal limits. MRI brain did reveal multiple tubers in the cerebral hemispheres located in convolutions in subependymal region projecting into the ventricular cavity producing a candle dripping appearance (Fig.3&4). There was no evidence of hydrocephalous. The echocardiogram and ultrasonographic examination of abdomen were also within normal limits. EEG did reveal hypsarrthymic pattern.

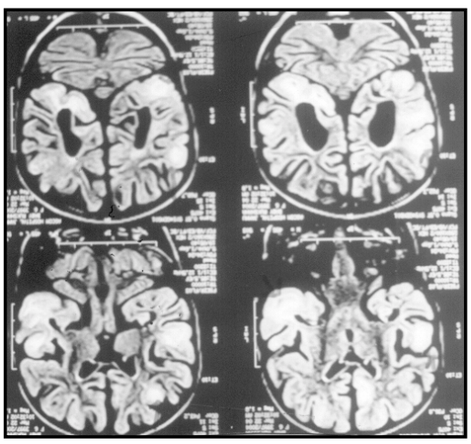

Fig. 3. MRI showing multiple tubers in the cereberal hemisphere

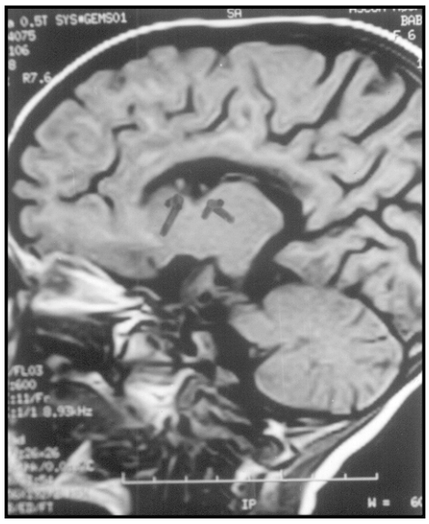

Fig. 4. Tubers projecting into ventricular cavity producing a candle dripping apperance

In view of clinical details, MRI Brain and EEG, the child was diagnosed as a case of Tuberous sclerosis. The child was advised to improve the nutritional status. She was prescribed sodium valproate, vigabatrin and haematinics. The seizures declined drastically.

Discussion

The first clear description of tuberous sclerosis (TS) was given by Bourneville in 1880 (3). It is inherited as an autosomal dominant trait. The TS gene is located on chromosome 9q34 (TSC) and 16 p 13 (TSC_2), but at least half of the cases are sporadic owing to new mutations (1,2,6). The younger the patient presents with symptoms of TS, the greater is the likelihood of mental retardation.

The child usually presents with myoclonic jerks. Other seizures including atypical absence spells, tonic and akinetic may also occur (2). Careful examination of the skin on trunk and extremities show hypopigmented skin lesions that have been likened to an ashleaf in more than 90% of cases in early childhood. (1-4). Our child too have classical skin lesions on face and trunk. Sebaceous adenomas develop between 4 and 6 years of age (1-3). They appear as tiny red nodules over the nose and cheeks are sometimes confused with acne. They

may enlarge, coalesce and assure a fleshy appearance. Our child did not have such lesions. Though he did have a roughened, raised lesion with an orange peel consistency on right side of forehead. This type of skin lesion called as shagreen patch is most often located primarily in the lumbosacral region (1,2). Subungual and periungal fibromas (koenentumor) arise from stratum lucidum of the fingers and toes during adolescence in 50 % of cases with TS (2). Retinal lesion in TS are of two types mulberry tumors that arise from nerve head and hakomas which are retinal hamartomas of astriocytic origin (1-7). Our child did not have these finding on detailed fundus examination. Approximately 50% of children with TS have rhabdomyoms of the heart. These may be numerous or located at the apex of the left ventricle, and although they can cause congestive heart failure and arrythmias, they tend to slowly resolve spontaneously (1). The echocardiogram done in our case was normal. The kidneys in most patients are involved by hamartomas or polycystic disease, resulting in hematuria, pain and in some cases renal failure. Generalised cystic or fibrous pulmonary changes in the lung may lead to spontaneous pneumothorax (1). In our case ultrosongraphy of kidneys and X-ray chest were within normal limits.We managed the child symptomatically. Besides maintaining the nutritional status, hematinics, anticonvulsants in form of sodium valproate and vigabatrin were prescribed. The frequency of seizures declined.

To conclude any child who presents with seizures must be looked for skin lesions which can give some clue towards the causation of seizures.

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