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Case Report

Tuberous Sclerosis in a Child

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Abstract

A female child presented with the complaints of seizure and learning difficulties. On examination various types of skin lesion were found, including adenoma sebacum, hypomelanic macules ("ash leaf spots"), Shagreen patches. On investigation, CT scan of brain showed subepedymal nodule in walls of ventricle. B scan of eyes revealed multiple tiny nodular lesions in posterior chamber of eyes called astrocytic hamartomas (or "phakomas"). Antiepileptic drugs started, now patient is seizure free.

Key words: Tuberous sclerosis, seizure, Ash leaf spots, shagreen patches, subependymal nodule.

Introduction

Tuberous sclerosis or tuberous sclerosis complex (TSC) is a rare, multi-system genetic disease that causes non-malignant tumors to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs, and skin. A combination of symptoms may include seizures, developmental delay, behavioral problems, learning difficulties, skin abnormalities, lung and kidney disease. TSC is caused by a mutation of either of two genes, TSC1 and TSC2, which encode for the proteins hamartin and tuberin respectively. These proteins act as tumor growth suppressors, agents that regulate cell proliferation and differentiation.

The name, composed of the Latin tuber (swelling) and the Greek skleros (hard), refers to the pathological finding of thick, firm and pale gyri, called "tubers," in the brains of patients postmortem. These tubers were first described by Désiré-Magloire Bourneville in 1880; the cortical manifestations may sometimes still be known by the eponym Bourneville's disease.

The Case

A 5 years old female child presented in paediatric OPD with the complaints of seizure and learning difficulties since 1.5 yrs. On detail history, type of seizure was tonic and clonic, for 10–15 minutes duration and not associated with loss of consiousness. School performance of the patient had gradually decreased since the illness had started, developmental milestone was normal. There was no history of head trauma, ear discharge, vomiting and speech abnormality. On examination various types of skin lesion were found, including adenoma sebacum which is a reddish, multiple, papulonodular lesion on the nose and cheek, butterfly in distribution. Hypomelanic macules ("ash leaf spots"): White or lighter patches of skin that appear over the face, arm and chest. Shagreen patches: Areas of thick leathery skin that are dimpled like an orange peel, found in front of the right ear. There was ptosis in left eye. Except seizure and skin lesion, the child did not have any symptoms pertaining to heart, kidney and lungs.

Investigation reports revealed, hemoglobin 10 gm%, TLC 9000 cells/ cumm. CT scan of brain showed subepedymal nodule in walls of ventricle, no ventriculomegaly. B scan of eyes revealed multiple tiny nodular lesions in posterior chamber of eyes called astrocytic hamartomas (or "phakomas"). Urine examination report was normal. X ray chest- No abnormality detected in lung field, cardiac size was normal. Abdominal USG was normal.

Examination findings and investigation reports were suggestive of tuberous sclerosis. Symptomatic treatment with tab carbamazepine was started for epilepsy; however the patient was not put on specific medicine for tuberous sclerosis because it is still controversial. Follow-up after 2 weeks patient was seizure free but other feature were same.

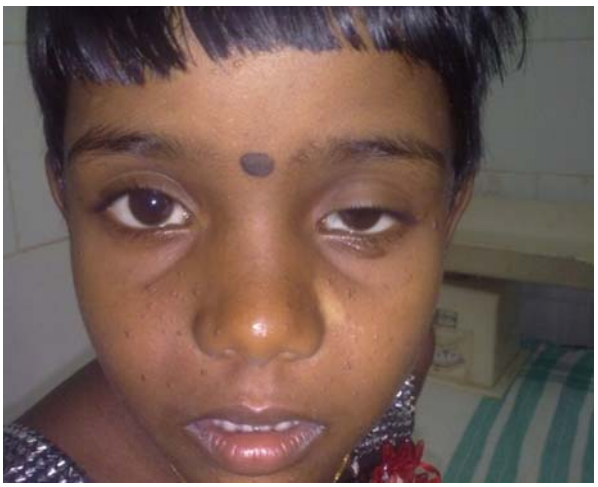

Fig 1: Photograph of a child showing Adenoma sebacum

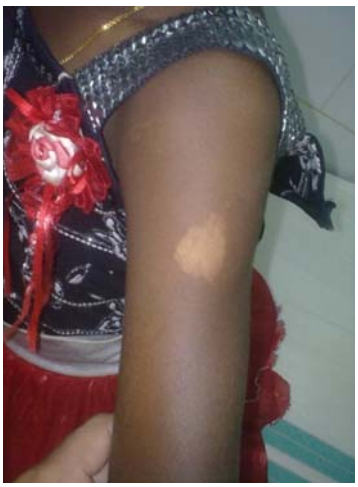

Fig 3: Showing Hypomelanic macules ("ash leaf spots")

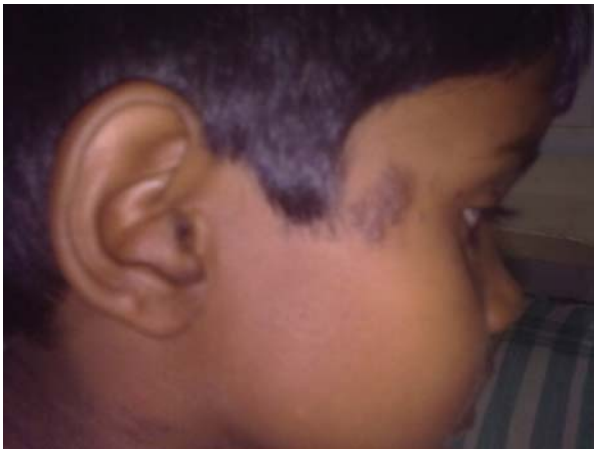

Fig 2: Photograph showing leathery skin patch " Shagreen patch"

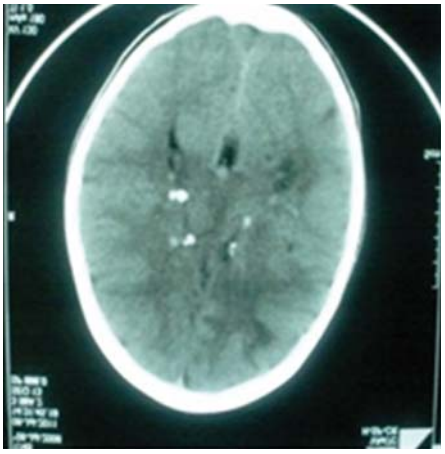

Fig 4: The CT Scan head showing subependymal nodules

Discussion

The physical manifestations of tuberous sclerosis are due to the formation of hamartia (malformed tissue such as the cortical tubers), hamartomas (benign growths such as facial angiofibroma and subependymal nodules) and, very rarely, cancerous hamartoblastomas. The effect of these on the brain leads to neurological symptoms such as seizures, developmental delay, and behavioral problems. About 50% of people with TSC have learning difficulties ranging from mild to significant[1]. My patient also had history of seizure and learning difficulties.

Three types of brain tumours may be associated with TSC: (i) Giant cell astrocytoma: grows and blocks the CSF flow leading to dilatation of ventricles causing headache and vomiting (ii) Cortical tubers: after which the disease is named. (iii) Sub-ependymal nodules: form in the walls of ventricles. Classic intracranial manifestations of tuberous sclerosis include subependymal nodules and cortical/subcortical tubers[2]. CT finding of my patient shows sub ependymal nodules in walls of ventricles.

The kidneys in most patients are involved by hamartomas or polycystic disease, resulting in hematuria, pain, and, in some cases, renal failure[3-4]. In our patient no such symptoms were found.

Patients with TSC can develop progressive replacement of the lung parenchyma with multiple cysts[3]. My patient did not have any feature related to lung abnormality.

Approximately 50% of children with TS have rhabdomyomas of the heart, which may be detected in a fetus at risk by an echocardiogram. The rhabdomyomas may be numerous or located at the apex of the left ventricle, and although they can cause congestive heart failure and arrhythmias, they tend to slowly resolve spontaneously[(3,5)]. Echocardiography of my patient was normal.

Some form of dermatological sign will be present in 96% of individuals with TSC. Most cause no problems

but are helpful in diagnosis. Some cases may cause disfigurement, necessitating treatment. The most common skin abnormalities include:

Facial angiofibromas ("adenoma sebaceum")[4,5]: A rash of reddish spots or bumps, which appear on the nose and cheeks in a butterfly distribution. They consist of blood vessels and fibrous tissue. This socially embarrassing rash starts to appear during childhood and can be removed using dermabrasion or laser treatment. Similar lesions with similar distribution are present in my patient also.

Periungual fibromas: Also known as Koenen's tumors, these are small fleshy tumors that grow around and under the toenails or fingernails and may need to be surgically removed if they enlarge or cause bleeding. These are very rare in childhood but common by middle age. These lesion were not present in the patient.

Hypomelanic macules ("ash leaf spots")[4,5]: White or lighter patches of skin that may appear anywhere on the body and are caused by a lack of melanin. These are usually the only visible sign of TSC at birth. In fair-skinned individuals a Wood's lamp (ultraviolet light) may be required to see them. In my patient these lesions are present in face, right arm and chest.

Shagreen patches[4]: Areas of thick leathery skin that are dimpled like an orange peel, usually found on the lower back or nape of the neck. This lesion is present in front of right ear in our case.

Retinal lesions, called astrocytic hamartomas (or "phakomas"), which appear as a greyish or yellowish-white lesion in the back of the globe on the ophthalmic examination[4,6]. In my patient also, multiple tiny nodular lesions are present in posterior chamber of eye. Astrocytic hamartomas (or "phakomas") is not a usual finding in investigation of TS, It is present in < 15% of the cases.

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