Tuberous sclerosis: a case report

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A case of tuberous sclerosis (TS) with classic triad of seizures, mental deficiency and angiofibromas is presented. The child also presented with self-mutilating behavior, which resulted in the fracture of her anterior teeth. A comprehensive treatment was rendered to the patient under general anesthesia and a mouth guard was given to prevent further trauma to the teeth.
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INTRODUCTION

Tuberous sclerosis (TS) also called tuberous sclerosis complex (TSC) is a dominant autosomal disease with an incidence of 1 in 20,000 to 30,000 individuals.[1] It is related to alterations in 9 and 16 chromosomes.[2] In TS, the classic triad consists of seizures, mental deficiency and angiofibromas.[3]

Bourneville, in 1880, gave the first detailed description of the neurological symptoms and cerebral pathology in three patients and coined the term tuberous sclerosis.[4] However, as early as 1862, Von Recklinghausen described cardiac myomas and sclerotic lesions of the brain in an infant.[5] Historical aspects of the disorder have been especially well reviewed by Gomez,[6] and Morgan and Wolfort.[7] TS is also know as Bourneville-Pringle Syndrome, Epiloia, and Adenoma sebaceum syndrome.[3]

Tuberous sclerosis complex (TSC) is an autosomal dominant genetic disorder characterized by a variety of organ system changes. The name is derived from the characteristic changes that occur in the brain, called tubers. Over time, these tuberous growths become hard and sclerotic and may calcify. Until recently, most of the proliferations and tumors associated with the disease were thought to be hamartomas.[3] Recent molecular biological analysis has revealed that these are clonal processes and true neoplastic growth. TS is characterized by a potential for hamartomatous growth in multiple organs and has a broad range of expression.[3]

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The current diagnostic criteria divide the disease into definite TSC and possible TSC:[3]

- Definite TSC: Either 2 major features or 1 major feature and 2 minor features.
- Possible TSC: Either 1 major features or 2 or more minor features.

Major features[3]

1. Facial angiofibromas or forehead plaque
2. Nontraumatic ungula or periungual fibroma
3. Hypomelanotic macules (more than 3)
4. Shagreen patch (Connective tissue nevus)
5. Multiple renal nodular hamartomas
6. Subependymal nodule
7. Subependymal giant cell astrocytoma
8. Cardiac rhabdomyoma, single or multiple
9. Lymphangiomyomatosis
10. Renal angiomyolipoma

Minor features[3]

1. Mutiple randomly distributed pits in dental enamel
2. Hamartomatous rectal polyps
3. Bone cysts
4. Cerebral white matter migration lines-may be diagnosed radiologically.
5. Gingival fibromas
6. Nonrenal hamartomas
7. Retinal achromatic patch
8. Confetti skin lesions
9. Multiple renal cysts.

Oral manifestations[8]

Oral manifestations of TS are:

1. Fibromas of the gum, the tongue and the palate
2. Papillomas
3. Odontogenic tumors and alveolar thickening
4. The pathgnomonic lesion in teeth is enamel hypoplasia characterized by small cavities of variable size. They are smaller and imperceptible in deciduous teeth and frequently require staining to be identified.[1]

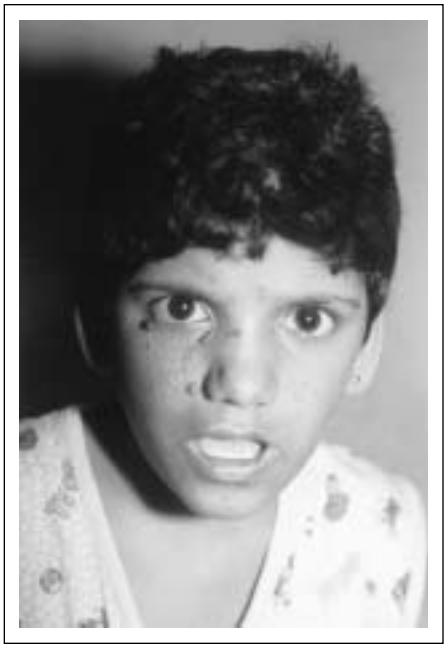
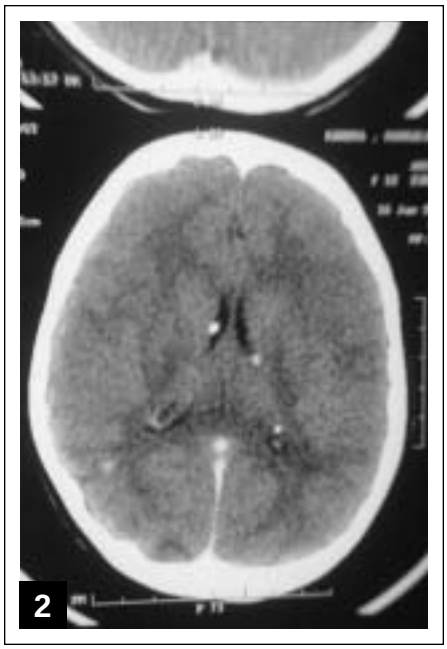
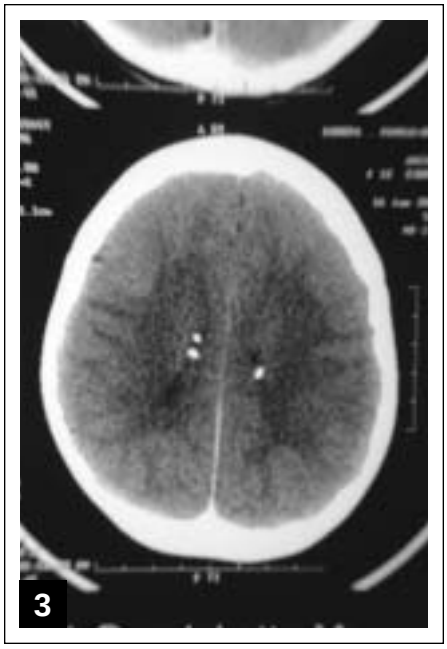


Figure 1. Photograph of face of patient with angiofibromas, Hypomelanotic macules; Ash leaf spots and Shagreen patches, aggressiveness is seen in her eyes.





Figures 2 and 3. Photograph of CT Scan showing "calcific nodules" in the brain.

CASE REPORT

A 12-year female patient reported to Department of Pedodontics and Preventive Childrens' Dentistry with a chief complaint of broken teeth in upper front region. On clinical examination the child presented with a combinations of symptoms including: 1. developmental delay, 2. behaviour problem, 3. seizures, 4. skin abnormality, and 5. mental retardation.

Behavioral assessment of the patient found the patient to be aggressive, to have intense rage and with self–mutilating behavior, which resulted in the fracture of her anterior teeth. Self-care activity was poor. Mother gave the history of destructive behavior towards her siblings.

Past medical history

Past medical history revealed that the patient was epileptic and was on medication up to 10 years of life. At present, patient had discontinued the medications. Mother reported that the child had delayed milestones of speech, growth, and cognitive development. Prenatal and family histories were non-contributory.

Psychiatric assessment

Psychiatric assessment revealed that patient is mentally retarded having the mental age of one.

Extra oral examination

Extra oral examination revealed that, the child had a normal gait. Characteristic angiofibromas was evident on face and also on other parts of the body. Hypomelanotic macules; ash leaf spots and shagreen patches were also seen on the face (Figure 1).

Intra oral examination

On intra oral examination, patient was suffering from poor oral hygiene. Multiple randomly distributed enamel hypoplastic pits were seen on the labial surface of the anterior teeth. Chronic generalized marginal gingivitis was found. Patient was also suffering from the multiple carious teeth in the posterior region. Ellis' Class III fracture was seen (Figure 1).

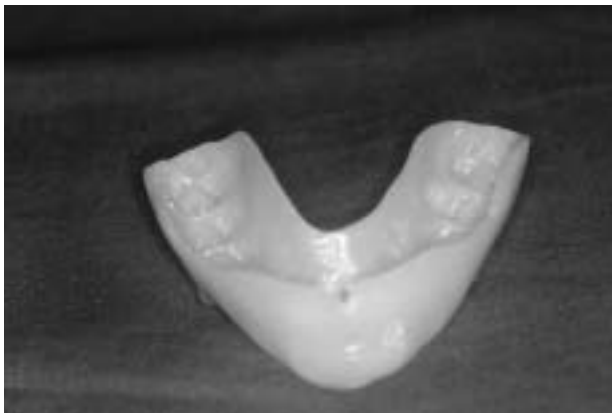


Figure 4. Photograph of mouthgaurd given to the patient to prevent further trauma to the anterior teeth.

Radiographic examination

The radiographs were taken by premedicating the child with IV diazepam, as patient was highly uncooperative. Orthopantomograph was taken to evaluate the dental status. CT scan was taken for the confirmation of the disease. Evidence of calcific nodules seen in the subependymal region and parenchymal calcifications were also noted in the CT scan (Figures 2 and 3).

Treatment

A comprehensive treatment was rendered to the patient under GA, as patient was highly uncooperative, after taking consent from the pediatrician and anesthetists. As a preventive measure against the self-mutilating behavior of the child, mouth guard was given to prevent further trauma to the teeth (Figure 4). Parents were counseled for the home care measures.

The patient was recalled after one month for review and was found to be asymptomatic.

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