

# Tuberous sclerosis with oral manifestations: A rare case report

SPS Sodhi, Ramandeep Singh Dang<sup>1</sup>, Gursimrat Brar

Department of Oral and Maxillofacial Surgery, Dasmesh Institute of Research and Dental Sciences, <sup>1</sup>Department of Neurosurgery, Guru Gobind Singh Medical College and Hospital, Faridkot, Punjab, India

## ABSTRACT

Tuberous sclerosis complex (TSC) is a neurocutaneous syndrome, inherited as an autosomal dominant trait with a high incidence of sporadic cases and protean clinical expression, with an incidence of prevalence between 1 in 10,000 and 1 in 170,000. The cardinal features of TSC are skin lesions, convulsive seizures, and mental retardation. We report a sporadically occurring case of definite TSC in a young female who presented with oral and cutaneous manifestations without mental retardation or history of convulsive seizures, which to the best of our knowledge has not been reported so far.

**Key words:** Convulsive seizures, mental retardation, neurocutaneous syndrome, tuberous sclerosis complex

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## INTRODUCTION

Tuberous sclerosis complex (TSC) is an autosomal dominant neurocutaneous syndrome with a high incidence of sporadic cases.<sup>[1]</sup> It was first described in 1862 by von Recklinghausen and later was more completely elucidated by Bourneville, Pringle, and Vogt (Rushton, 1955).<sup>[2]</sup> It is a disorder of cellular differentiation and proliferation leading to the development of benign tumors such as neurofibromas and angiofibromas and with high penetrance but considerable variability in the expression.<sup>[3]</sup>

The genes thought to be responsible for tuberous sclerosis are located on chromosomes 9q and 16p.<sup>[4]</sup> The birth prevalence is as high as 1 in 5800 persons.<sup>[5,6]</sup> The cardinal features of TSC are skin lesions, convulsive seizures, and

mental retardation.<sup>[1]</sup> Ninety percent of tuberous sclerosis patients develop various seizure disorders, and 60% develop measurable mental retardation.<sup>[5,6]</sup>

Tuberous sclerosis is characterized by variability and expressivity of clinical manifestations and is often age-related [Table 1].<sup>[1]</sup> School-aged children usually present with adenoma sebaceum, developmental delay, learning disability/retardation, and seizures.<sup>[1]</sup> Orofacial manifestations include fibrous hyperplasia, hemangiomas, facial asymmetry, bifid uvula, cleft lip and palate, macroglossia, high arched palate, delayed eruption, diastemas, and enamel defects.<sup>[7]</sup>

## CASE REPORT

An 18-year-old female reported to the Department of Oral and Maxillofacial Surgery at Dasmesh Institute of Research and Dental Sciences, Faridkot with the chief complaint of painless swelling on the left side of face since 8 years and reddish discoloration of skin on the same

Address for correspondence: Dr. Ramandeep Singh Dang, Department of Neurosurgery and Surgical Critical Care, Guru Gobind Singh Medical College and Hospital, Sadiq Road, Faridkot, Punjab India. E-mail: ramandang@yahoo.com

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**Table 1: Diagnostic criteria for TSC**

## Primary features

- Facial angiofibromas
- Multiple ungual fibromas
- Cortical tuber
- Subependymal nodule or giant cell astrocytoma
- Multiple calcified subependymal nodules protruding into the ventricle
- Multiple retinal astrocytomas

## Secondary features

- Affected first-degree relative
- Cardiac rhabdomyoma
- Other retinal hamartoma or achromic patch
- Cerebral tubers
- Noncalcified subependymal nodules
- Shagreen patch
- Forehead plaque
- Pulmonary lymphangiomyomatosis
- Renal angiomyolipoma
- Renal cysts

## Tertiary features

- Hypomelanotic macules
- “Confetti” skin lesions
- Renal cysts
- Randomly distributed enamel pits in deciduous or permanent teeth
- Hamartomatous rectal polyps
- Bone cysts
- Pulmonary lymphangiomyomatosis
- Cerebral white matter “migration tracts” or heterotopias
- Gingival fibromas
- Hamartoma of other organs
- Infantile spasms

## Diagnostic categories

- Definite TSC: One primary feature, two secondary features, or one secondary plus two tertiary features
- Probable TSC: Either one secondary plus one tertiary feature or three tertiary features
- Suspect TSC: Either one secondary feature or two tertiary features

TSC: Tuberous sclerosis complex

side of face and neck since birth with no positive family history [Figure 1].

History of repeated excisions of gingival growth was present since patient was 4 years old. On general physical examination, the patient was well-oriented to time, place, and person; moderately built with normal gait and IQ. She had a nodular swelling on the middle finger of the left hand. Magnetic resonance imaging (MRI) brain and carotid angiograph were suggestive of cortical and sub-cortical hyperintensities with gyral calcification; common with (c/w) tuberous sclerosis [Figure 2]. Orthopantomography showed a multilocular mixed radiolucent-radiopaque lesion with incomplete septae involving the left mandibular region eroding the posterior ramus of mandible causing an altered shape of the left mandibular ramus, condyle, and coronoid process [Figure 3].



Figure 1: Painless swelling on the left side of face



Figure 2: Magnetic resonance imaging brain and carotid angiograph



Figure 3: Orthopantomography showing a multilocular mixed radiolucent-radiopaque lesion

As our case showed only skin lesions, our line of treatment is regular age-dependent screening to assess the patient's behavioral, cognitive, and neurological functions. MRI of the brain will be performed every year to assess the risk factors for developing astrocytomas. In this case, the prognosis seems good, as the patient presented with only oral and cutaneous manifestations without any mental retardation or convulsive seizures.

## DISCUSSION

The diagnosis of tuberous sclerosis has evolved from only clinical observations to the use of anatomicopathological studies to that using molecular biology.<sup>[8]</sup> The disease develops as an abnormal growth of ectodermic and mesodermic cells producing benign tumors extending to areas of the head, heart, brain, and kidneys. The term epiloia (epilepsy, low intelligence, and adenoma sebaceum) was proposed by Campbell and Sherlock.<sup>[9]</sup> This designation may be of some use in describing the disease although all the three signs are rarely present. The classic triad of epiloia is seen in only 30% of affected individuals.<sup>[8]</sup>

We report this case as a rare phenomenon because of its sporadic occurrence in a young female who presented with oral and cutaneous manifestations without mental retardation or history of convulsive seizures, which to the best of our knowledge has not been reported so far. Patients with TSC must adopt measures for careful oral and dental hygiene, with regular visits to the dentist, in order to eliminate potential irritative factors and ensure the early diagnosis of any possible lesions.<sup>[3]</sup> The cerebral manifestations and renal complications of tuberous sclerosis exert an important influence on patient prognosis.<sup>[10]</sup> The diagnosis and management of these patients vary depending on the specific systemic presentation of the disease. A dentist's role in early diagnosis of such lesion is helpful in the management of these lesions. Regular follow-up is helpful to prevent any systemic involvement if any in future. The need for a detailed medical history is therefore always beneficial for the proper management of these patients.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other

clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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### Conflicts of interest

There are no conflicts of interest.

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