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## Peripheral cemento-ossifying fibroma associated with TUBEROUS SCLEROSIS

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### ABSTRACT

Tuberous sclerosis complex (TSC) was a rare autosomal dominant neurocutaneous disease characterized by benign tumors affecting various body systems, skin changes, neurological disorders, and multi organ development of hamartomas leading to morbidity and death. Intraoral fibromas, gingival hyperplasia and enamel hypoplasia or enamel pits are the most common oral manifestations. Those patient management always involves a multidisciplinary approach from various fields. Here we present a case study of 35 years old female patient with tuberous sclerosis complex characteristic clinical, radiological, and histological features.

**Keywords:** Tuberous sclerosis, Fibromas, Hamartoma.

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## INTRODUCTION

A rare autosomal dominant condition, first identified in 1880 by Désiré Magloire Bourneville, is tuberous sclerosis complex (TSC) or Bourneville disease. A rare, multisystem genetic disorder that affects multiple systems and exhibits symptoms of central nervous system, cardiovascular, renal, respiratory, ocular, and dermatological manifestations. Peripheral cemento-ossifying fibroma(PCOF) is a rare lesion of variable forms. It is characterized as a well-marked and often encapsulated lesion consisting of fibrous tissue that contains variable amounts of bone-like mineralized material (ossifying fibroma), cementum (cemento-ossifying fibroma) or both.

### **Clinical manifestation:**

The term “Tuberous sclerosis” derives from the tuber or potato-like nodules in the brain, which calcifies with age, and progress to become hard or sclerotic. Most of the TSC patients develop various disorders in the first year of life. It typically affects the central nervous system and leads to seizures, delays in development, behavioral issues. This lesion's pathogenesis is unknown and is thought to derive from the periosteal and periodontal membrane. However, there is no evidence to support this connection and their occurrence remains unexplained in areas distant from the periodontal ligament.

PCOF constitutes 3.1% of all oral tumours, and 9.6% of all gingival lesions. It will happen at any age but peaks in the second to third decades. The average age is around 28 years and common in females.

The predilection of females was recorded to be as high as 5:112. Clinically, PCOFs are sessile or pedunculated, usually ulcerating (or) manifesting as a colour similar to that of the gingiva surrounding them. PCOF should be surgically excised for confirmation of diagnosis and should be subject to histopathological examination.

### **CASE REPORT:**

A 35 year old female patient came to our department with the chief complaint of swelling in her right upper back tooth region for past 2 years. The patient noticed the growth for the past 2 years, which was initially smaller in size and gradually increased in size, Asymptomatic. The patient is a known epileptic and not on any medications. On general physical examination, the face was Asymmetrical with, the appearance of red papules on face especially in nasolabial folds, cheek and chin suggestive of Adenoma sebaceum[Figure 1]. Sessile and firm nodular growths were also observed in the lower extremities suggestive of periungual fibroma or Koenen tumours [Figure 2]. Subungual fibroma was seen in the little finger (Phalanx) [Figure 3] and retinal hamartomas were

also present. Competent lips and right submandibular lymph nodes were palpable



**Figure 1: Adenoma sebaceum – upper part of face**



**Figure 2: Periungual fibroma or Koenen tumors**



**Figure 3: Subungual fibroma in thumb fingers.**

On intra oral examination, a well-defined sessile, discrete, nodular growth, fibrous in consistency measuring approximately 3x4 cm in size was evident extending from 16-18 region. Hard tissue examination revealed generalized attrition, erosion, and dental pits were seen with poor oral hygiene.

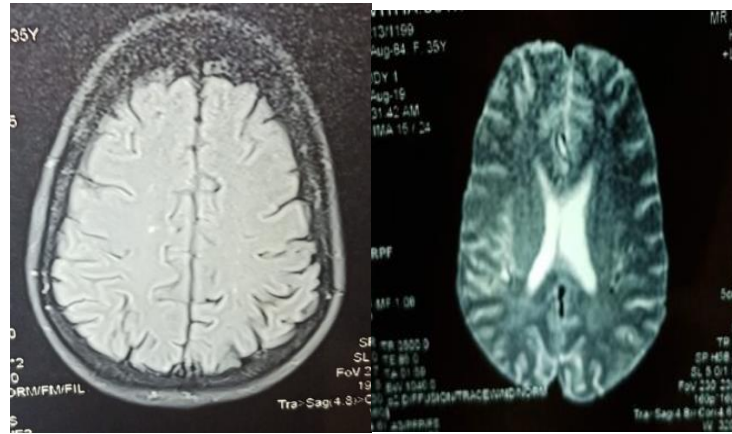


**Figure 4: Sessile nodular growth evident in 16-18 region (buccal and palatal aspect)**

The patient was subjected to radiological, haematological and histopathological investigations were carried out, evident as revealed by the medical record. Orthopantomogram revealed evidence of bone destruction in relation to 16 and 18 with multiple proximal caries and root stumps. Generalized horizontal bone loss was evident in the upper arch [Figure 5]. Ultrasonography of the abdomen revealed simple cortical renal cyst of size 21x 16mm seen in the left mid pole. Right and left ovary was little larger and it shows multiple small cysts with heterogeneous echo pattern. Mild hepatomegaly (fatty infiltration of liver), Congestive liver and minimal Ascites, minimal pericardial effusion was seen. Magnetic resonance imaging (MRI) showed T2/ FLAIR hyperintensity noted in right frontal gyrus – cortical tuber, Small sub centimetric T1/T2 hypointense foci noted in posterior lobe which shows blooming- calcified granuloma [Figure 6].



**Figure 5: Orthopantomogram and IOPA shows evidence of bone destruction in relation to 16 and 18**



**Figure 6: Magnetic Resonance Imaging shows blooming calcified granuloma.**



**Figure 7: Chest X-Ray**

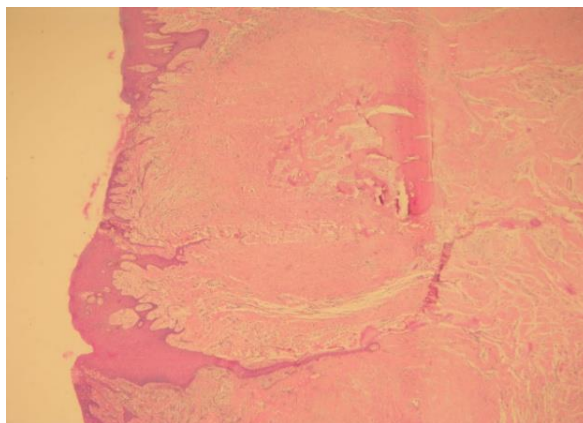
Subsequently, excisional biopsy of gingival growth in relation to 16 and 18 was carried out. Under Local anesthesia, using 3.0 black silk the lesion was completely encircled and ligated and lesion was completely excised using scalpel Following which tooth 16 and 18 were extracted. Periodontal dressing was given and patient was prescribed regular medications.



**Figure 8: Lesion was engaged with suture material and Excisional biopsy done in relation to 16 and 18**



**Figure 9: Excision on palatal aspect and linear measurements were made.**



**Figure 10: Histopathological image**



**Figure 11: 3 Months follow up – Post OP**

Histopathology section of the specimen revealed, the surface lined with hyperplastic non keratinized stratified squamous epithelium. Highly cellular connective tissue exhibiting plump fibroblast in moderately fibrous connective tissue trabeculae of metaplastic bone formation and few calcifications are noted. The peripheral area shows lymphocytic infiltrations.[Figure 10]

#### DISCUSSION:

TSC is characterized by the development of tumours in brain, skin, retina, and viscera. The diagnosis of TSC is based on the identification of hamartomas in more than one system.<sup>[1]</sup> The

abnormal genes is located at one of two locations, the long arm of chromosome 9 (9q34) and the short arm of chromosome 16 (16p 13.3) designated as TSC2 (encoding tuberlin)<sup>[1]</sup>

The presence of two major characteristics or one major and two minor characteristics was considered adequate for a definitive diagnosis. The key characteristics include (a) hypomelanotic macules (=3), a minimum diameter of 5 mm, (b) facial angio fibroma (=3) or fibrous cephalic plaque, (c) ungula fibromas (= 2), and (d) shagreen patches. The minor characteristics include: (a) "confetti" skin lesions (b) dental enamel pits (=3), and (c) intraoral fibromas (=2).<sup>[2]</sup> TSC's most common oral manifestations are fibromas, gingival hyperplasia, and enamel hypoplasia (in almost 100 percent of these patients are associated with increased caries risk).

Other features include the rectal polyps of hamartoma, non-renal hamartomas, numerous renal and bone cysts, high arched palate, bifid uvula, cleft palate, delayed dental eruption, and diastema. Our case had three major features facial angiofibroma, koenen tumour and subependymal nodules and two minor features-gingival fibromas and enamel hypoplasia.<sup>[3]</sup> Such patients also have multidisciplinary treatment including the neurosurgeon, neurologist, nephrologist, pulmonologist, cardiologist, ophthalmologist, and genetic counsellor. Surgery including dermabrasion and treatment with lasers can be useful for treating skin lesions. Intervention programs including special schooling and occupational therapy may benefit individuals with special needs and developmental concern.<sup>[1]</sup> Modalities of treatment for some of the TSC symptoms are currently in progress. The use of topical 0.1 per cent rapamycin on facial angiofibroma has been demonstrated in recent studies. The use of mTOR inhibitors in the regression of different growths of hamartoma is a new way of handling TSC. These patients must adopt measures for careful oral and dental hygiene with regular visits to the dental surgeons to eliminate the potential irritant factors and ensure the early diagnosis of any possible lesions.<sup>[1]</sup> The prognosis of TSC depends on multiple system involvement. About a quarter of severely affected infants is thought to die before the age, of 10% and 75% before 25 years.

#### CONCLUSION:

Thus, proper detection of clinical oral features such as dental enamel pits and fibroma will help to obtain an early diagnosis of this disease and eventually to facilitate effective screening, treatment and genetic counseling.

#### REFERENCES:

1. S. Sarkar, T. Khaitan, R. Sinha, and A. Kabiraj, "Tuberous sclerosis complex: A case report," *Contemp. Clin. Dent.*, vol. 7, no. 2, pp. 236–239, 2016.
2. H. Northrup et al., "Tuberous sclerosis complex diagnostic criteria update:

Recommendations of the 2012 international tuberous sclerosis complex consensus conference,” *Pediatr. Neurol.*, vol. 49, no. 4, pp. 243–254, 2013.

3. K. Harutunian, R. Figueiredo, and C. Gay-Escoda, “Tuberous sclerosis complex with oral manifestations: A case report and literature review,” *Med. Oral Patol. Oral Cir. Bucal*, vol. 16, no. 4, 2011.
4. Webb DW, Clarke A, Fryer A, Osborne JP. The cutaneous features of tuberous sclerosis: a population study. *Br J Dermatol* 1996;135:1–5. [PubMed: 8776349]
5. Stirrups DR, Inglis J. Tuberous sclerosis with nonhydantoin gingival hyperplasia. Report of a case. *Oral Surg Oral Med Oral Pathol*1980;49:211–213. [PubMed: 6928303]
6. Thomas D, Rapley J, Strathman R, Parker R. Tuberous sclerosis with gingival overgrowth. *J Periodontol*1992;63:713– 717. [PubMed: 1507052]
7. Roach ES, Gomez MR, Northrup H. Tuberous sclerosis complex consensus conference: revised clinical diagnostic criteria. *J Child Neurol*1998;13:624–628. [PubMed: 9881533]

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