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Tuberous Sclerosis

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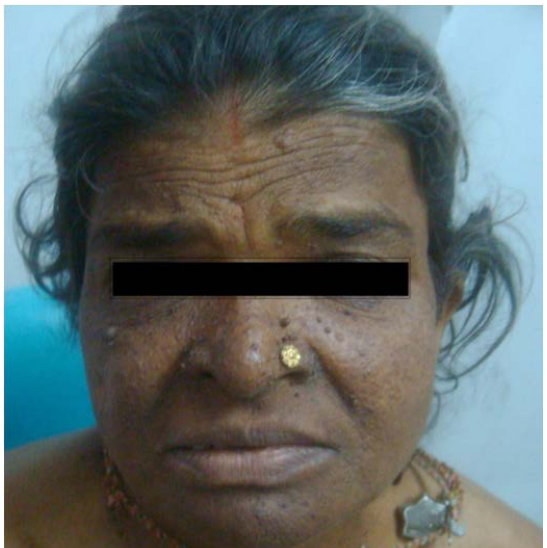
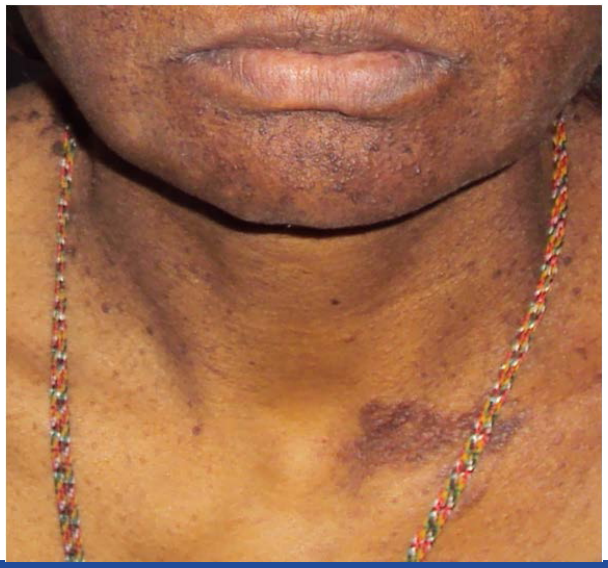
Tuberous Sclerosis (also known as Bournville-Pringle syndrome) is a rare disorder, usually linked to a triad of conditions comprising epilepsy, mental retardation, and angiofibromas, as well as to oral and skin manifestations, though all the three signs are rarely present. A partial form of the condition is usually observed. The disease develops as an abnormal growth of ectodermic and mesodermic cells producing benign tumors that extend to areas of the head, heart, brain, and kidneys [1]. We present to you a case report of a patient who came to the department with a chief complaint of foul smell from the mouth and was later diagnosed as having tuberous sclerosis on the basis of her clinical features and further imaging studies.

A 45-year-old woman reported to the Department of Oral Medicine and Radiology, with a chief complaint of foul smell from the mouth since six months. On general examination, she was anaemic in appearance [Table/Fig-1] and was having small multiple brown fleshy overgrowths of variable diameter present over the entire face extending upto the neck [Table/Fig-1,2]. Among these a few flesh coloured orange-peel connective tissue naevi of varying sizes were present on the anterior portion of clavicular region on left side (Shagreen patch) [Table/Fig-2]. There were smooth, firm, flesh-coloured lumps emerging from the nail folds of hand [Table/Fig-3] and feet [Table/Fig-4] (Periungual fibromas). A solitary fleshy overgrowth was present on the right toe which was pinkish white in colour [Table/Fig-4]. On intraoral examination, patient was having a poor oral hygiene. There was presence of multiple pin headed nodules on both sides of the buccal mucosa [Table/Fig-5]. On dorsal surface of tongue there was presence of grayish pigmentation, multiple fissures and coated tongue [Table/Fig-6].

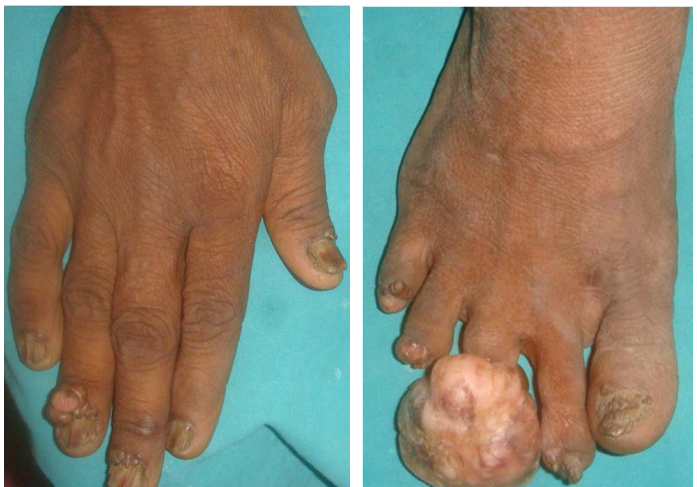
A panoramic radiograph was taken to evaluate any bony changes. The radiograph revealed presence of multiple radiolucencies in the mandible along the mandibular canal [Table/Fig-7]. Skull AP and Lateral views were taken, showing no alterations. Blood investigations were done, which showed microcytic hypochromic anemia with Hb value of 4.2 gm/dl. The WBC count was 5300/cumm, Platelet count was 3.16 lacs/cmm, Serum creatinine was 1.62 mg/dl, and PCV was 17.1%. The rest of the values were within normal limits. The patient was referred to a physician for



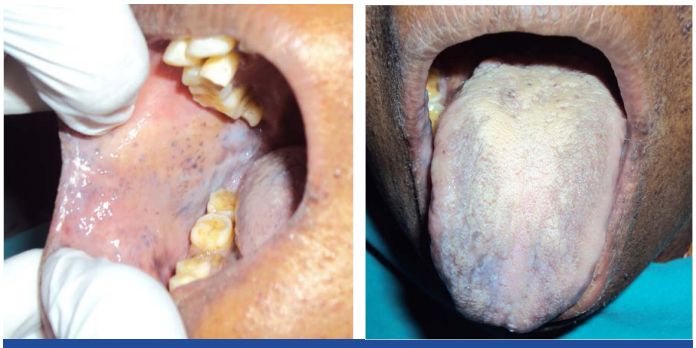
[Table/Fig-1]: Clinical picture showing small multiple brown fleshy overgrowths of variable diameter present over the entire face extending upto the neck (Facial Angiofibromas)



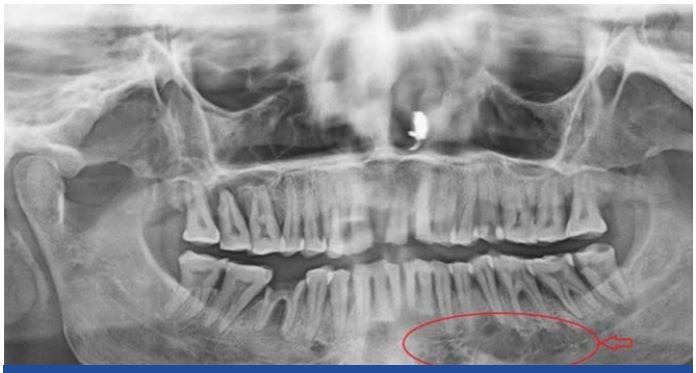
[Table/Fig-2]: Flesh coloured orange-peel connective tissue naevi of variable diameter, present in relation to left side of anterior portion of clavicular region (Shagreen patch)



[Table/Fig-3]: Smooth, firm, flesh-coloured lumps (Periungual fibromas) emerging from the nail folds of hand **[Table/Fig-4]:** Smooth, firm, flesh-coloured lumps (Periungual fibromas) emerging from the nail folds of feet

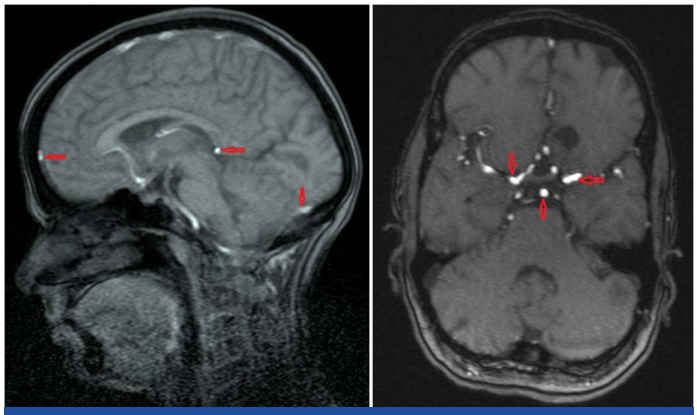


[Table/Fig-5]: Multiple pin headed nodules present bilaterally on the buccal mucosa; **[Table/Fig-6]:** Grayish pigmentation.on dorsal surface of tongue



[Table/Fig-7]: Panoramic radiograph showing presence of multiple radiolucencies in the mandible along the mandibular canal

further evaluation and to rule out the presence of cysts and tumors in other organs. The physician conducted an ultrasonographic evaluation, in which no sonographic abnormalities were detected. In order to evaluate any neurological manifestations, CT and MRI scan was advised. The CT scan of brain revealed presence of subependymal nodules which were present near the walls of the cerebral ventricles. MRI scans revealed presence of multiple cortical tubercles and giant cell histiocytomas [Table/Fig-8,9]. In order to check the malignant potential of the growth on the second finger of right foot, an excisional biopsy was performed, which confirmed the growth to be fibrosarcoma. Multiple Endocrine Neoplasia (MEN 1) was considered as the differential diagnosis, but as all the findings present in the patient were fulfilling the criterias of tuberous sclerosis, final diagnosis of tuberous sclerosis was made.



[Table/Fig-8]: CT scan of brain showing presence of subependymal nodules
[Table/Fig-9]: MRI scans revealed presence of multiple cortical tubercles and giant cell histiocytomas

Patient was admitted and blood transfusion was done. The Hb increased from 4.2 gm/dl to 10.1 gm/dl while PCV value increased from 17.1% to 34%. The patient refused for any treatment and was discharged from the hospital. In dental management, oral prophylaxis was done.

Elaborate review of the published cases revealed that most of the patients diagnosed with Tuberous Sclerosis had seizures, epileptic spasms, mental deficiency and cardiac involvement [2-4]. While in our case, we came across dermatological findings as the primary symptom of the disease which led to further diagnosis. Seizures, epileptic spasms, mental deficiency and cardiac involvement were not seen in our case.

REFERENCES

[1] Cutando A, Gil JA, López J. Oral health management implications in patients with tuberous sclerosis. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod.* 2000;89:430-35.
[2] Monteiro T, Garrido C, Pina S, Chorao R, Carrilho I, Figueiroa S, et al. Tuberous sclerosis: clinical characteristics and their relationship to genotype/phenotype. *An Pediatr (Barc)*. 2014;81(5):289-96.
[3] Prabhu S, Mahesh KP. Tuberous sclerosis with oral angiofibroma: case report. *Br J Oral Maxillofac Surg.* 2010;48(3):205-07.
[4] Mettin RR, Merkenschlager A, Bernhard MK, Elix H, Hirsch W, Kiess W, et al. Wide spectrum of clinical manifestations in children with tuberous sclerosis complex--follow-up of 20 children. *Brain Dev.* 2014;36(4):306-14.

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