International Journal of Medicine, 11 (2) (2023) 20-23

International Journal of Medicine

Website: www.sciencepubco.com/index.php/IJM

Case Report

Tuberous Sclerosis or Bourneville-pringle disease: A rare case report

Harsh Kumar Veshar [1] *, Veer Bahadur Singh [2], Mayank Shrivastava [3], Ibrahim Khan [4], Kapil Singh Meena [5], Sourabh Soni [1], Amit Kant [1] Saranshi Singh [6]

[1] *Senior Resident, JLN Medical College Ajmer Rajasthan*

[2] *Senior Professor, JLN Medical College Ajmer Rajasthan*

[3] *Associate Professor, JLN Medical College Ajmer Rajasthan*

[4] *Assistant Professor, JLN Medical College Ajmer Rajasthan*

[5] *Assistant Professor, GMC Sirohi Rajasthan*

[6] *Junior Resident, JLN Medical College Ajmer Rajasthan*

**Corresponding author E-mail: hveshar@gmail.com*

Abstract

Introduction: Tuberous sclerosis or Bourneville's disease is rare autosomal dominant neurocutaneous. Which is characterized by Skin changes, neurological issues, and the development of hamartomas in several organs that cause morbidity and mortality. The gingival hyperplasia, fibromas, hypopigmented lesion's and enamel hypoplasia are the most typical features of presentation. The case report emphasises on the crucial clinical characteristics, radiographic characteristics, and laboratory abnormalities of Tuberous Sclerosis assisting in the early detection and treatment of such a rare illness.

Case presentation: In this report, we present a case of tuberous sclerosis in a 25-year- old female who displayed abnormal body movement along with a hypopigmented macule, ash leaf spots, periungual fibroma, gum hyperplasia, and hematuria with pain in the bilateral flank.

Conclusion: Our aim is to raise awareness for tuberous sclerosis by using this instance to highlight the disease's rarity and the difficulty in identifying and treating cases like this.

Keywords: *Tuberous Sclerosis; Neurofibromatosis; Skin Diseases; Fibroma; Hamartoma.*

1. Introduction

Von Recklinghausen originally identified the rare autosomal dominant condition known as tuberous sclerosis complex (TSC) in 1862. Based on the pathologic characteristics of the sclerotic tubers discovered during the postmortem examination of individuals with epilepsy and mental retardation, Bourneville created the word "sclerose tubereuse" in 1880, which is now known as Bourneville's illness.[1] Specifically, the skin, brain, eye, kidney, and heart have hamartoma formation in this case. Angiofibroma's, periungual fibromas, shagreen patches, and ash leaf white macules are the typical skin lesions that are typically, though notalways, associated with epilepsy and mental retardation.[2] Sherlock came up with the acronym EPILOIA to encapsulate this condition because it is clinically distinguished by thetriad of epilepsy (EPI), intellectual disability (LOI), and adenoma sebaceum (A).[3] TSC is around one third as prevalent as neurofibromatosis type 1 and is expected to occur in 1 in 10,000 live births.[4] It is a multisystemic illness that only manifests in late childhood, making early diagnosis in infancy impractical.[5].

2. Case report

A 25-year-old female presented JLN hospital Ajmer(North-West Rajasthan), casualty department with chief complaint of Abnormal body movement since two days. On first day she had two attacks lasting for around 1-2minutes, on second day she had an attack of abnormal body movement which lasted around 8-10minutes, which were associated with urinary incontinence, tongue bite & frothing from mouth, these all were suggestive of Generalized tonic clonic type of seizure for which she got admitted in hospital was treated with intravenous phenytoin loading dose followed by its maintenance. She is known case ofGTCS since 8 years, had been on antiepileptic medications but convulsions were under control. She had no other similar past history.

On initial examination she was Afebrile, drowsy, abdomen was soft non tender but there was lump in both side abdomen, blood pressure was 100/76mm of Hg respiratory rate 16/min bilateral lung air present, hearts sounds were normal no murmur heard, bilateral plantar flexor & bilateral pupil normal & reactive to light & no signs suggestive of any kind of focal neurological deficit.

On head to toe examination, patient had facial angiofibromas on forehead, around nasolabial folds, which were characteristic pattern reddish sessile nodular growth in butterfly shape around nasolabial fold, which were suggestive of Adenoma Sebacium. . On inspection of the oral cavity, upper gums had well-defined, hard nodular development. These gingival enlargements are most likely caused by antiepileptic drug toxicity (the causative drug is phenytoin), according to the clinical examination and history discussed above. The differential diagnosis of neurofibromas and gingival fibromas was carefully considered. Bothupper and lower limbs had similar, varying-sized sessile and hard nodular growths. Just lateral to the little finger nail on the left hand, a soft, compressible lesion was found that was a Köenen tumour or periungual fibroma. On the left side of the abdomen, close to the umbilicus, the right posterior trunk was seen to have a well-defined localised hypopigmentedlesion. Right lumbosacral region with an orange peel appearance suggestive of shagreen patch and well-defined roughened hypermelanotic patch on the right shoulder.

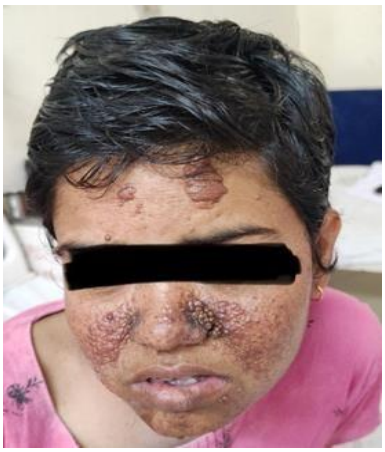

Fig. 1: Adenoma Sebaceum.

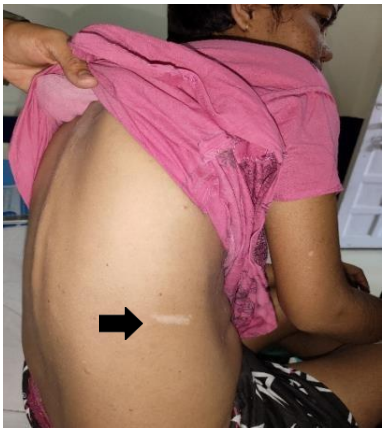

Fig. 2: Hypopigmented Ashleaf Macules.

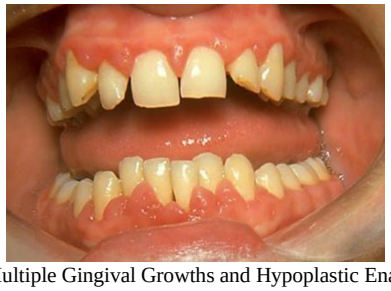

Fig. 3: Multiple Gingival Growths and Hypoplastic Enamel Pits.

After the physical examination, blood and radiological workups were done. A complete bloodcount and biochemistry investigation were found to be inside the normal limits. In Urine examinations, haematuria was seen with 34 RBCs /high power field. After this, a radiological examination was done, which revealed significant findings pertaining to our case.

On radiological examination chest orthopentogram was grossly normal. On abdomen ultrasonography it was suggestive of multiple bilateral hypoechoic lesion renal in the cortex of kidney. Other then this there was no abnormality in ultrasonography. In order to confirm these lesions, a CECT Abdomen scan was done, and the results revealed numerous small, hypodense lesions with fluid attenuation in the bilateral renal cortex, which are suggestive ofrenal angiomyolipoma and renal cortical cysts. MRI Brain was suggestive of Subependymal nodules & typical subependymal giant cell astrocytomas(SEGA).

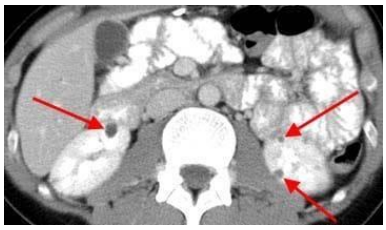

Fig. 4: CECT Abdomen Showing Bilateral Renal Angiomyolipoma.

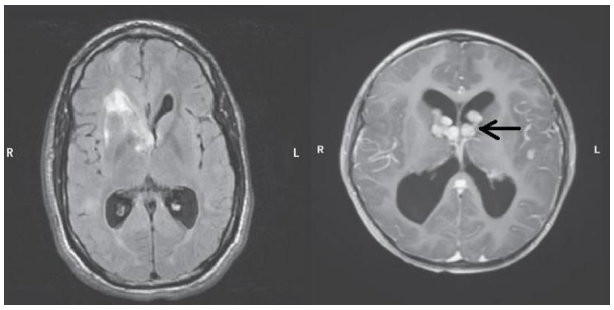

Fig. 5: Subependymal Nodules & Typical Subependymal Giant Cell Astrocytoma's (SEGA).

Table 1: Clinical Diagnostic Criteria for Tuberous Sclerosis Complex

Clinical Diagnostic Criteria For TSC

MAJOR FEATURES		MINOR FEATURES	
1	Hypomelanotic macules (≥3, at least 5mm)	1	"Confetti" skin lesions
2	Angiofibromas (≥3) or fibrous cephalic plaque	2	Dental enamel pits (≥3)
3	Ungual fibromas (≥2)	3	Intraoral fibromas (≥2)
4	Shagreen patch	4	Retinal achromic patch
5	Multiple retinal hamartomas	5	Multiple renal cysts
6	Cortical dysplasias (≥3)*	6	Nonrenal hamartomas
7	Subependymal nodules (≥2)		
8	Subependymal giant cell astrocytomas		
9	Cardiac rhabdomyoma		
10	Lymphangioleiomyomatosis (LAM)**		
11	Angiomyolipomas (≥2)**		

* Includes **tubers** and cerebral white matter **radial migration lines**.
** A combination of the two major clinical features LAM and angiomyolipomas without other features does not meet criteria for a Definite Diagnosis.

3. Discussion

Tuberous sclerosis (TS), also known as Bourneville disease, is a rare genetic disorder of autosomal-dominant inheritance with a prevalence ranging from one in 6,000 to one in 12,000. Both sexes and all ethnic groups can be affected. [6 - 8] It is characterized by theskin, brain, eye, kidney, and heart have hamartoma formation in this case. Facial Angiofibroma's, periungual fibromas, shagreen patches, and ash leaf white macules are the typical skin lesions that are generally present, though not always, associated with epilepsy and mental retardation. The clinical diagnostic criteria for TSC were updated, and a new classification scheme based on major and minor features was created, during the 1998 Tuberous Sclerosis Consensus Conference. For a conclusive diagnosis, the presence of two main traits or one major and two minor features were sufficient to make diagnosis of tuberoussclerosis.[9] Mutations in the tumor-suppressor gene TSC1 (encoding hamartin) or, more frequently, TSC2 (encoding tuberin), which result in a lack of mTOR pathway inhibition, are linked to the etiology of TS. As a result, hamartomas begin to develop in a variety of organs, such as the skin, lungs, heart, and kidneys. The brain also develops hamartomas, including cortical tubers, subependymal nodules, and subependymal giant cell astrocytoma's.
In our case there were 7 major & 1 minor criteria were present which are as followed hypomelanotic macules, facial angiofibroma's, periungual fibromas, shagreen patches, renal angiolipomas, typical subependymal giant cell astrocytoma's(SEGA), and sub-ependymal nodules. One minor criteria intraoral fibroma. Hence we make diagnosis of tuberous sclerosiscomplex.

4. Conclusion

Tuberous sclerosis is rare autosomal dominant neurocutaneous syndrome in which there is multiple system involvement therefore multi-disciplinary approach should be applied for itsmanagement.
One should include tuberous sclerosis in differential diagnosis once they cutaneous lesion associated with seizure. Genetic counselling should be offered to the parents, even though there is no sure shot genetic test due to its variability in gene expression. There are pharma-cotherapies but one should focus on increasing the quality of life to be lived by suchpatients.

References

[1] Dumitrescu D, Georgescu EF, Niculescu M, Dumitrescu CI, Mogoanta SS, Georgescu I. Tuberous sclerosis complex: Report of two intrafamilial cases,both in mother and daughter. *Rom J Morphol Embryol*. 2009;50:119–24. [PubMed] [Google Scholar].
[2] Tony Burns, Stephen, Breathnach Neil cox et al. Tuberous Sclerosis complex InRooks textbook of Dermatology, Volume 1 Massachusetts Blackwell science, 2004;355:1345-1356. https://doi.org/10.1002/9780470750520.

[3] Illahi Y, Tanveer S, Khurshid Pasha KA, Naeem A, Ali N. Tuberous sclerosis.Classical presentation in a male patient. *NMJ*. 2010;2:29–32. [Google Scholar].
[4] Midde ML, Saheb DM. Tuberous sclerosis complex – A case report. *SoutheastAsian J Case Rep Rew*. 2013;2:343–8. [Google Scholar].
[5] Shrestha S, Shrestha S, Ojha AR. Case report on tuberous sclerosis. *J Kathmandu Med Coll*. 2013;2:208–10. [Google Scholar]. https://doi.org/10.3126/jkmc.v2i4.11798.
[6] Tuberous Sclerosis Complex Board of Osaka University Hospital. Tuberous sclerosis complex: recent advances in manifestations and therapy. Wataya- Kaneda M, Uemura M, Fujita K, Hirata H, Osuga K, Kagitani-Shimono K, Nonomura N. https://doi.org/10.1111/iju.13390.
[7] Tuberous sclerosis. Curatolo P, Maria BL. *Handb Clin Neurol*. 2013;111:323–331. [PubMed] [Google Scholar]. https://doi.org/10.1016/B978-0-444-52891-9.00038-5.
[8] Tuberous sclerosis complex: a review. Leung AK, Robson WL. *J Pediatr Health Care*. 2007;21:108–114. [PubMed] [Google Scholar]. https://doi.org/10.1016/j.pedhc.2006.05.004.