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TUBEROUS SCLEROSIS IN FAMILY- A CASE REPORT

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ABSTRACT
Tuberous Sclerosis Complex is a genetically inherited rare disorder in autosomal dominant fashion. It is a multisystem disorder involving skin, brain, heart, kidneys, lungs and eyes, which becomes more apparent after late childhood, which is limiting the usefulness of early diagnosis in infancy. We report a case of tuberous sclerosis in 13-year male child and his father.

INTRODUCTION

Desiree- Magloire Bourneville was first described Tuberous sclerosis complex (TSC) or Bourneville's disease in 1880, as an autosomal dominant inheritance with the prevalence of one in 6000 live birth, which is affecting sexes, all races and ethnic groups [1,2]. This disease results from mutations in one of two genes, *TSC1* encoding hamartin or *TSC2* encoding tuberin, which plays an essential role in the regulation of cell proliferation and differentiation [3,4]. It is a multisystem disorder involving skin, brain, heart, kidneys, eyes and lungs, which becomes more apparent after late childhood, limiting the usefulness for early diagnosis in infancy.

CASE REPORT:

A13 year's old boy presented with complaints of multiple dark coloured skin lesions over the face for past 6 years. There are no history of seizures and mental retardation. His birth and developmental history were unremarkable. Patient's father also had similar complaints since childhood. Other family members did not have any skin lesions. There were no other systemic manifestations or symptoms.

His neurological examination was unremarkable. Dermatological examination revealed multiple papular lesions of dark brown coloured of about 5 -10 mm in size, seen on all over the body especially on nose, nasal bridge and extending to the cheek region in butterfly like distribution suggestive of facial angiofibromas ("adenoma sebaceum"). Thick leathery skin areas that are dimpled like an orange peel and pigmented areas seen on right back side of the patient suggestive of 'Shagreenpatches'.

Hypomelanotic white patches seen on lower back region, suggestive of 'ash leaf spots'. Raised, discolored areas on the forehead suggestive of forehead plaques.

Ophthalmological evaluation was normal. Electroencephalography (awake record) revealed no abnormalities. Ultrasonography of abdomen, chest x-ray and echocardiography were noncontributory. A provisional diagnosis of Tuberous Sclerosis Complex Syndrome was established. And radiological investigations were performed. Contrast CT Scan study of scalp was done and that report showed a solitary hypo dense area measuring about 5x5 mm in size seen in axial section**,** suggestive of **'**Subependymal calcification or nodules'.

Figure 1. Clinical photograph showing multiple pigmented papules and plagues over the face of both father and son.

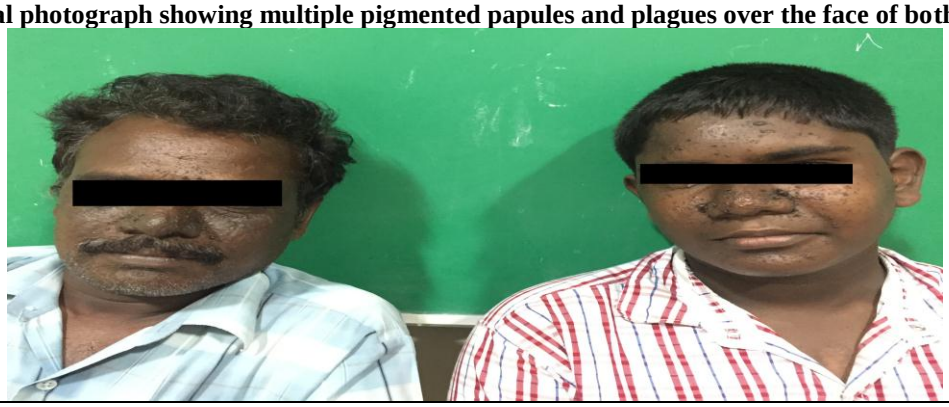


Figure 2. Clinical photograph of 13year old boy showing lesions involving nose, nasal bridge and extending to the cheek region in butterfly like distribution of facial angiofibromas.

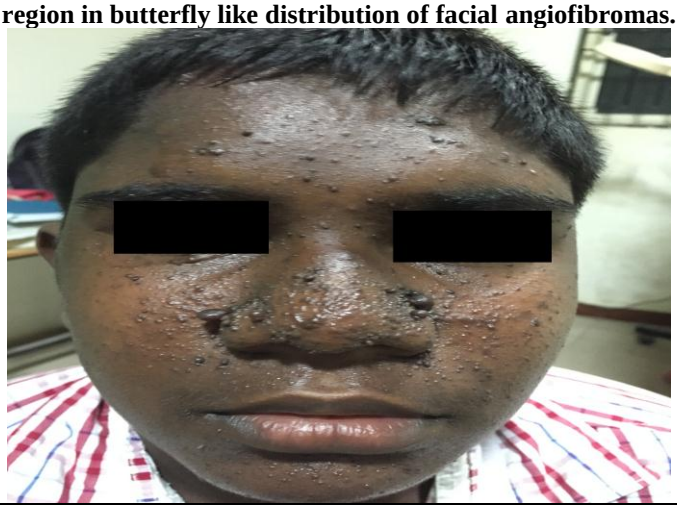
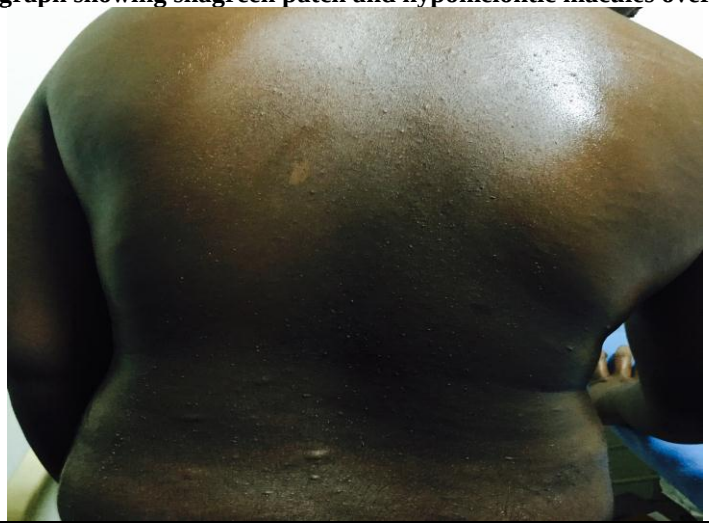


Figure 3. Clinical photograph showing shagreen patch and hypomelontic macules over the back of the patient.



DISCUSSION

Tuberous sclerosis complex (TSC) is a genetically inherited rare neuro-cutaneous disorder with heterogeneous presentation varying from severe mental retardation and incapacitating seizures to normal intelligence and an absence of seizure, often within the same family. It is due to inactivating the mutation in one of the two genes, TSC1 gene encoding hamartin, or TSC2 gene encoding tuberin.

The major neurological manifestations of tuberous sclerosis complex are epilepsy, autism, developmental delay and behavioral and psychiatric disorder. Epilepsy is present in about 80-90% of patient. TSC has dermatologic manifestations like Shagreen patch (20-30%), facial angiofibromas (75%), hypomelanotic macule (90%). Hypomelanotic macules are present at birth and almost all

lesions will appear within the first two years of life. Facial angiofibromas (adenoma sebaceum) are appearing at the preschool years over the malar area. The shagreen patch is present over the lumbosacral region characteristically present as an irregularly shaped raised lesion with the consistency of orange peel. Adolescent pediatric children may have cosmetic issues, so recent trial supports the use of topical 0.1% Rapamycin on facial angiofibromas. Diagnostic Criteria for TSC is as given in the table below. Definite TSC can be made when two major or one major plus two minor features are demonstrated.

Major and Minor Criteria of tuberous sclerosis complex

Major Criteria

1. Cortical tuber
2. Subependymal nodule
3. Facial angiofibromas or forehead plaque
4. Ungual or periungual fibroma (nontraumatic)
5. Hypomelanotic macules (>3)
6. Shagreen patch
7. Multiple retinal hamartomas
8. Cardiac rhabdomyoma
9. Renal angiomyolipoma
10. Pulmonary lymphangioleimyomatosis

Minor Criteria

1. Cerebral white matter migration lines
2. Multiple dental pits
3. Gingival fibromas
4. Bone cysts
5. Retinal achromatic patch
6. Confetti skin lesions
7. Nonrenal hamartomas
8. Multiple renal cysts
9. Hamaromatous rectal polyps

Treatment of angiofibromas over the face is challenging. Therapeutic options include cryosurgery, curettage, excision, chemical peels, dermabrasion, laser therapy (e.g. pulse dye laser or ablative fractional resurfacing), electrosurgery, podophyllin, 5-aminolevulinic acid photodynamic therapy (PDT). A variety of degrees of success and side effects have been reported [7]. Recently, rapamycin (sirolimus), either topical or systemic, has been proposed for the treatment of cutaneous angiofibromas.

CONCLUSION

Tuberous Sclerosis Complex is a lifelong condition; therefore individuals with TSC should be regularly monitored by an experienced clinician. TSC must be included in the differentials of children presenting with seizures, developmental delay, and mental retardation. Early diagnosis is very important for thorough clinical and radiological evaluation, continuous monitoring of symptoms, family planning, genetic counseling and reduction in morbidity and mortality rate.

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CONFLICT OF INTEREST:

The authors declare that they have no conflict of interest.

STATEMENT OF HUMAN AND ANIMAL RIGHTS

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

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