# Case Report

# A rare case of tuberous sclerosis: A case report

Ankit Sharma, Alka Bhambri, Syed Moiz Ahmed

Department of Pediatrics, Rohilkhand Medical College, Bareilly, Uttar Pradesh, India

**Corresponding Author:**
Syed Moiz Ahmed, Department of Pediatrics, Rohilkhand Medical College, Bareilly, Uttar Pradesh, India. Phone: +91-7060457131. E-mail: drmoiz2008@gmail.com

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Tuberous sclerosis is a rare multisystem syndromic disease that is autosomal dominant and involves multiple organs causing non-cancerous growth in the brain and other vital organs. The incidence of this disease is very low involving 1 in 10,000 of new births. Most of the features of this syndrome become evident in early stages of life. Here, with this article, we report a case of 14-year-old female patient with this rare disease.

**KEY WORDS:** Autosomal dominanat, hamartomas, tuberous sclerosis

## INTRODUCTION

Tuberous sclerosis (TSC) is a rare multisystem syndromic disease that is autosomal dominant and involves multiple organs causing non-cancerous growth in the brain and other vital organ system such as cardiac, pulmonary, kidneys, liver, and eyes. The incidence of this disease is very low involving 1 in 10,000 of new births.[1] It was in 1862, when von Recklinghausen first described the disease. This is classified under neurocutaneous disorders and is clinically characterized by the classical triad of epilepsy, intellectual disability, and adenoma sebaceum.[2] Here, this case report describes about a 14-year-old female with TSC.

## CASE REPORT

A 14-year-old female patient presented to the outpatient department of pediatrics with the chief complaint of fever, cough, and abdominal pain for 10 days. It was not associated with any significant medical history and family history was also found to be not significant.

On general physical examination, generalized paleness was present over the body with well-defined multiple sessile blackish nodules involving forehead, around the eyes, and nose along with cheeks region [Figure 1]. Various hypomelanotic macules were present over the body with thick leathery skin over the trunk and lower back. A well-defined roughened hypermelanotic patch was noted in the left shoulder girdle and the right lumbosacral region showing an orange peel appearance indicative of shagreen patch [Figure 2]. On recording her vitals, she was febrile with breathing rate at 22–24 breaths per minute. The patient was thoroughly evaluated both clinically and radiologically. On ophthalmologic examination, multiple astrocytoma and hemorrhages were seen in her retina. The ultrasonography (abdomen) report showed a corticomedullary thickness with altered echo pattern and multiple small high echogenic foci diffusedly seen in both kidneys which were suggestive of angiomyolipoma. The MRI brain [Figure 3] report showed multiple cortical and subcortical tubers, non-calcified sub-ependymal tubers, and bilateral hypointense lesion areas of sclerosis. A provisional diagnosis of TSC was made keeping in mind all these above mentioned clinical and radiological features.

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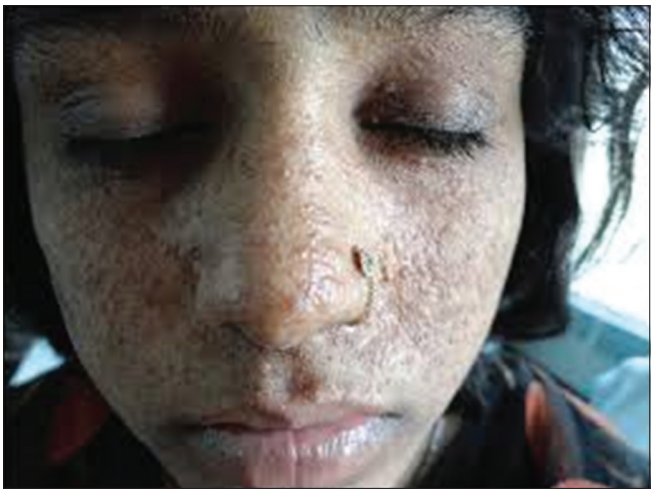

**Figure 1:** Adenoma sebacum

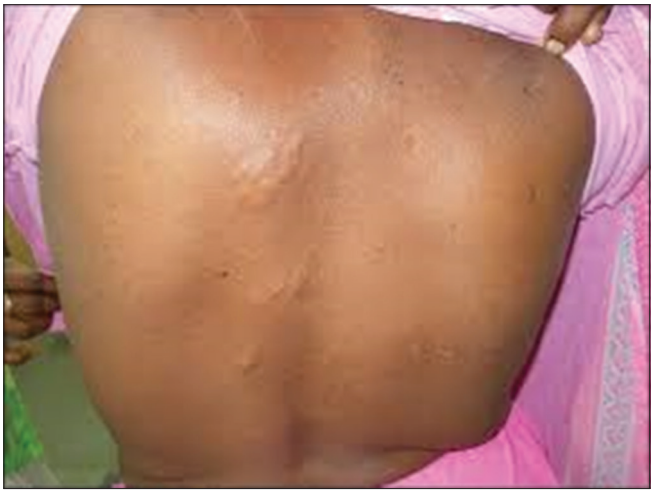

**Figure 2:** Shagreen patch

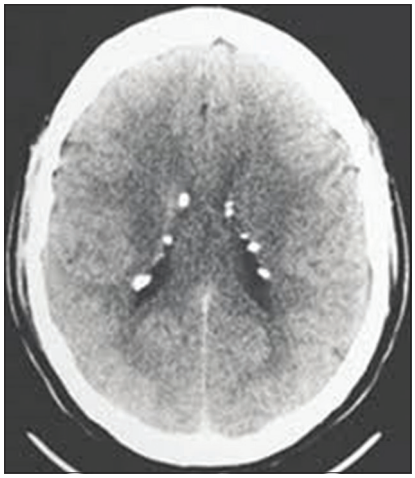

**Figure 3:** MRI finding

## DISCUSSION

The typical characteristic of TSC is the development of unusual tumor-like growths (hamartomas) in multiple vital organs of the body. The term "tuberous sclerosis" was coined due to the presence of multiple sclerotic masses scattered throughout the cerebrum.[3] TSC is an autosomal dominant disorder with variable expressivity but nearly complete penetrance. The abnormal genes designated as TSC 1 which are encoding hamartin are located at the long arm of chromosome number 9 (9q34) whereas, TSC 2 encoding tuberin is located at the short arm of chromosome number 16 (16p 13.3).[4]

The management protocol of these patients is multidisciplinary which involves participation of the neurosurgeon along with neurologist, nephrologist, cardiologist, ophthalmologist, odontologist, pulmonologist, as well as a genetic counselor. The management of skin lesions, include dermabrasion, surgical excision, electrocautery, and laser, are also found to be very useful. Intervention programs such as including special schooling due to their poor mental development and occupational therapy are essential for individuals of TSC requiring special needs as there is a persistent concern among parents regarding development of their child.[5]

The prognosis of TSC is variable as well as multifactorial. It depends on the number and severity of organs involved. In individuals diagnosed late in life and manifesting with only few cutaneous signs, then in such cases, prognosis may depend on development of internal tumors and cerebral calcifications. It has been found that about a quarter infants who are severely affected with the condition usually die before the attainment of 10 years of age and 75% succumb before attaining age of 25 years.[6,7]

These problems which are related to the brain development and dermatological outgrowths affect not only the emotional state of child but also in long run affect their social interaction and routine daily activities.[8] Hence, it is the responsibility and also the duty of the concerned medical team to follow these children and provides necessary psychological support apart from clinical aspect of this disease with a view to help decrease the social morbidities associated with the disease.

## CONCLUSION

TSC is an autosomal dominant disease affecting multiple organ system of the body and may be associated with non-cancerous growth involving brain as well as other vital organs. The seriousness of the disease can be controlled by its early detection and appropriate intervention to reduce the manifestations of serious complications. The overall clinical and radiological features confirmed the diagnosis of TSC. The patient was started with I.V. antibiotics, blood transfusion was given and referred to ophthalmologist, dermatologist, and surgeon.

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