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A CASE REPORT OF TUBEROUS SCLEROSIS COMPLEX

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ABSTRACT

Tuberous sclerosis complex (TSC) is a rare genetic disorder inherited as an autosomal dominant pattern with the prevalence of one in 6000 live birth, affecting both sexes and all ethnic groups. Tuberous sclerosis complex is characterized by the growth of numerous benign tumors in many parts of the body, including the brain, heart, lungs, eyes, kidneys, skin and other organs, leading to significant health problems like seizures, intellectual disability, autism or developmental delay .Tuberous sclerosis has no cure, but treatment such as medical, educational, and occupational therapy can help in relieving symptoms. Here we present a case of a young female patient with tuberous sclerosis with various clinical and radiological features who exhibited multiple hamartomas of various organ systems.

Key words: TSC, autosomal dominant, tumors, intellectual disability, autism

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INTRODUCTION

Tuberous Sclerosis or Tuberous Sclerosis Complex (TSC) is an autosomal dominant disorder characterized by the growth of numerous benign tumours in many parts of the body including the brain, heart, lungs, eyes, kidneys, skin and other organs. TSC is caused by mutations of two genes, TSC1 and TSC2, which encode for the proteins hamartin and tuberin respectively. These proteins act as tumour growth suppressors which are involved in cell proliferation and differentiation.[1] The estimated prevalence of TSC in general population is one in 95,136 and those <6 years of age is one in 14,608.[2] Genetic studies have shown that two thirds the disease results from a new dominant mutation either in the TSC1 gene on chromosome 9q343 or the TSC2 gene on chromosome 16p13, with the latter accounting for an estimated 78% of cases and do not have any affected parents.[3] About 80-90% of patients present with seizures in early first year of life which varies from subtle focal seizures, infantile spasms, to generalized tonic clonicseizures.[4] Definite TSC is diagnosed when either 2 major features (out of a total of 11) or one major feature with 2 minor features (out of a total of 9) are present. However approximately 50% of patients who fulfil the diagnostic criteria have normal intellectual ability

and 15% among them remain free from seizures.[5] Here we present a case of a young female with background history of seizures and multiple hamartomas involving various organs diagnosed as TSC using 2012 TSC diagnostic criteria.[6]

CASE REPORT

A 30 year old female, presented to the outpatient department of KIMS hospital with the history of fever since 4-5 days with no other localising symptoms. She was a known case of seizure disorder since 8 years of age receiving oral medications for seizures. On physical examination, there were multiple nodular skin lesions over face - adenoma sebaceum (fig.1), depigmented spots over back-ash leaf macules, nails showed periungal fibromas- kenyuontumour (fig.2) and a big nodular lesion over the left side of the scalp (4×5 cm) was seen (fig. 3).

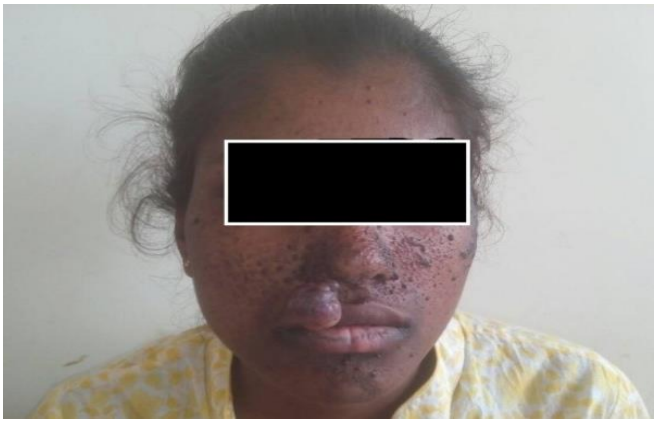

Figure 1: Multiple nodular skin lesions over face-adenoma sebaceum

Figure 2: Periungal fibroma- kenyuon tumor

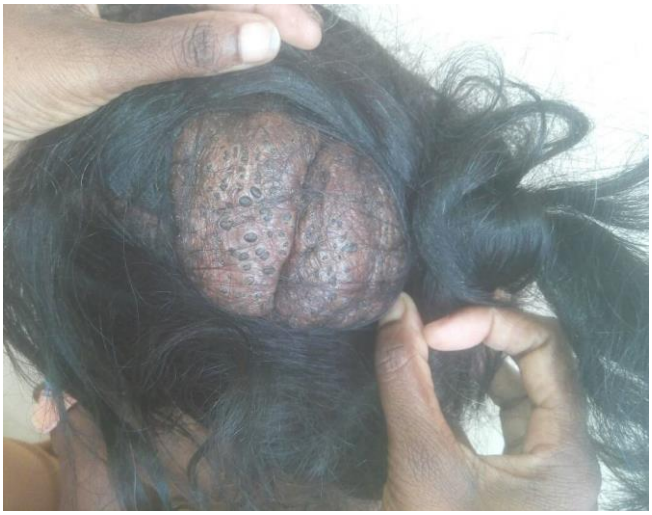

Figure 3: Nodular lesion on scalp

Investigations:

Complete blood picture showed Hb: 8.5, total count 3000, platelet count of 65000 and peripheral smear showed pancytopenia. Other investigations like liver function tests, renal function tests, serum electrolytes remained normal. USG abdomen showed multiple renal cysts, with 2 angiomyolipomas (size 1cm × 2cm).

Visual examination: Vision of 6/12 not improving with the pin hole. Fundoscopy showed multiple white hypo-pigmented lesions suggestive of non calcified retinal astrocytic hamartoma (fig.4).

CT brain showed sub ependymal tumours, (size of about 1× 2cm). Biopsy of the lesion over the scalp was taken which showed fibrous tissues (hamartoma) on histopathological examination.

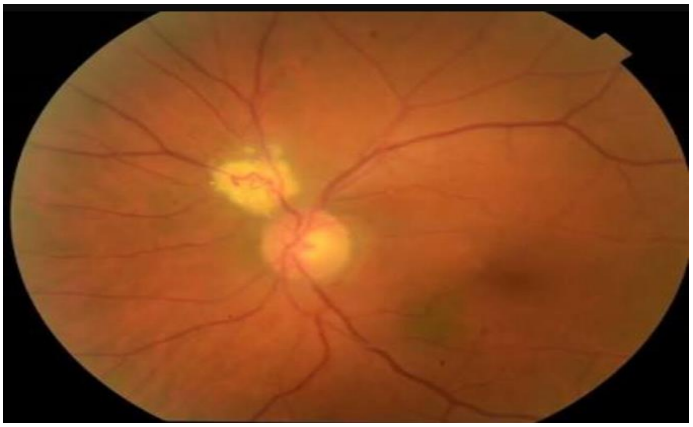

Figure 4: Non calcified retinal astrocytic hamartoma

The International Tuberous Sclerosis Complex Consensus Conference held in 2012, came with a new diagnostic criteria for diagnosis of tuberous sclerosis.[6] Definite diagnosis includes two major features or one major feature with 2 minor features Possible diagnosis includes either one major feature or 2 minor features. According to the above mentioned criteria our patient presented with more than 2 major criteria and was considered as TSC.

Major criteria

1. Hypomelanotic macules (3) at least 5-mm diameter
2. Angiofibromas (3) or fibrous cephalic plaque
3. Ungual fibromas (2)
4. Shagreen patch
5. Multiple retinal hamartomas
6. Cortical dysplasias
7. Subependymal nodules
8. Subependymal giant cell astrocytoma
9. Cardiac rhabdomyoma
10. Lymphangioleiomyomatosis
11. Angiomyolipomas (>2)

Minor criteria

1. "Confetti" skin lesions
2. Dental enamel pits (>3)
3. Intraoral fibromas (>2)
4. Retinal achromic patch
5. Multiple renal cysts
6. Nonrenal hamartomas

Based on the above clicinal findings and investigations, a final diagnosis of tuberous sclerosis was confirmed.

TSC is a lifelong condition and there is no definitive treatment for the disease. Patient was started on antiepileptics and was on continous monitoring of symptoms. Family planning and genetic counseling was done.

DISCUSSION

Tuberous Sclerosis is an important genetic disorder, which affects the patient and the family in various ways.[7] Various research projects are being done around the globe, to study more about genes involved and also the possible treatment strategies. Now, due to ongoing research, understanding of pathogenesis of disease, multiple drug therapies are available for certain manifestations of the disease.[8] With available medications, symptomatic control can be done. Children affected with tuberous scerosis can be offered schooling and occupational therapy.TSC presents with varied ophthalmic features. The retinal lesions more commonly include astrocytic hamartomas and non retinal lesions include coloboma, angoifibroma of the eyelid and papilledema (secondary to hydrocephalus). [9] Different surgeries such as dermabrasion and laser treatment can be offered to patients with skin lesions.[10]

CONCLUSION

Early diagnosis is very important. Radiology is important in diagnosis and detecting complex nature of the disease. After detailed clinical and radiological evaluation, monitoring of symptoms, family planning, and genetic counselling can decrease the rate of morbidity and mortality.

Declaration of Patient Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information

to be reported in the journal. The patient(s) understand(s) that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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