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Research Article

ROLE OF AN OPHTHAMOLOGIST IN A CASE OF TUBEROUS SCLEROSIS

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ABSTRACT

Tuberous sclerosis is a multi-systemic disorder with characteristic involvement of the eye. Often, the ophthalmological manifestations are either underdiagnosed or diagnosed at a stage when the eye cannot be saved. Here is one such case where the retinal involvement is detected at an early in a teenage boy.

INTRODUCTION

Tuberous sclerosis (TS) is a systemic disease which is characterized by hamartomas in multiple organs such as the skin, brain, heart, kidney, lungs and eyes. It was first described by Bourneville in 1880. In 1908, Vogt described the classic triad of epilepsy, mental retardation, and adenoma sebaceum (now known asangiofibromatosis). Patients with TS show mutations of the TSC1 and TSC2 genes, which encode for hamartin and tuberin respectively. Although this is an autosomal dominant hereditary disease, 60- 70% of all cases result from spontaneous mutations[1, 2]. The prevalence of TSC ranges from 1:6,000 to 1:10,000, and the diagnosis is usually established between 4-10 years of age or during puberty[3].

Van der Hoeve, in 1921, recognised ocular involvement as part of the same disorder. Uni- or bilateral retinal astrocytic hamartomas are present in approximately 50% of cases with tuberous sclerosis.

Case

A 14 year old boy presented with complaints of seizures. On examination, he was found to have mental retardation, epilepsy and facial angiofibromas, i.e the classic Vogt triad. Complete ophthalmic evaluation, CT and MRI of brain and ultrasound of abdomen were done.

Facial examination of the patient revealed multiple dark brown coloured, papular lesions of about 4 -10 mm over the face on nose, nasal bridge and the cheek region in butterfly like distribution suggestive of facial angiofibromas (adenoma sebaceum) [Figure 1].

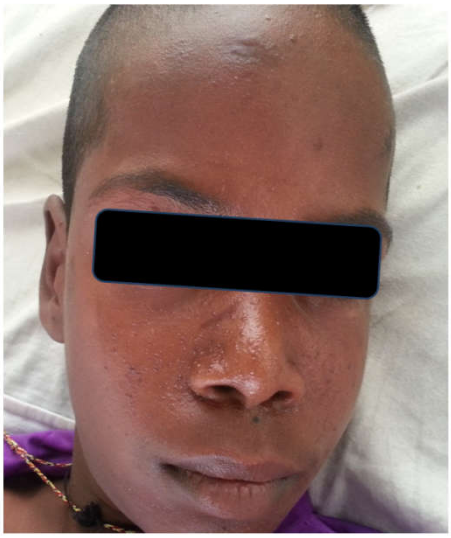

Figure 1 Facial examination of the patient revealed multiple dark brown coloured, papular lesions of about 5 -10 mm size, seen over the face on nose, nasal bridge and extending to the cheek region in butterfly like distribution suggestive of facial angiofibromas "adenoma sebaceum"

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- Visual acuity of both eyes was 6/6.
- Posterior segment evaluation showed multiple astrocytomas and hypopigmented patches in the retina of type III. [Figure 2]

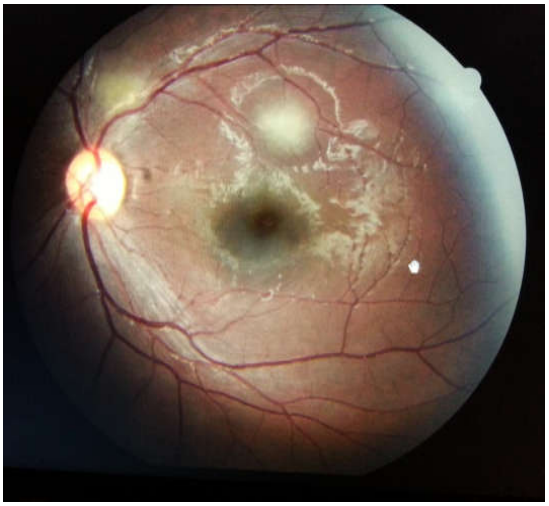

Figure 2 Posterior segment evaluation showed multiple astrocytomas and hypopigmented patches in the retina of type III.

- CT and MRI of brain showed subependymal calcified nodules and multiple cortical tubers.
- Ultrasound of abdomen showed polycystic kidneys.

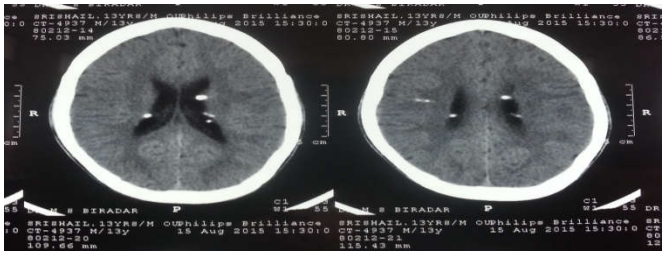

Figure 3 CT scan showed multiple hyperdense lesions which were suggestive of 'Subependymal calcification or nodules

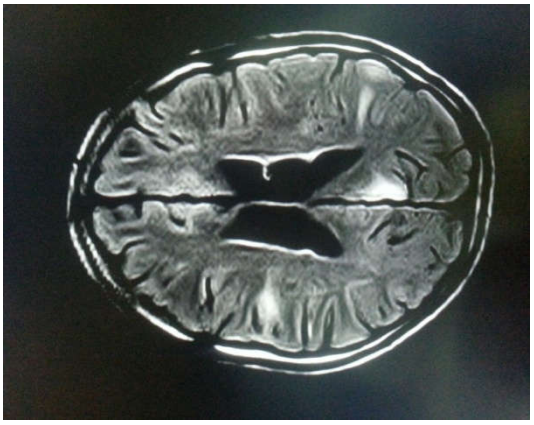

Figure 4 MRI of brain revealed multiple cortical tubers

DISCUSSION

Ophthalmic manifestations of TSC are divided into retinal and non-retinal. Retinal hamartomasaca be of the following types:(i) Retinal hamartomas can be non-calcified, grey, translucent lesion; (ii) the elevated, multinodular, calcified, opaque lesion resembling mulberries; and (iii) a transitional lesion which has morphological features of both of the previous. Other retinal changes seen are in the form of retinal pigmentary disturbances, either hyperpigmented areas (likely congenital retinal pigment epithelium hypertrophy) or "punched out" hypopigmented areas at the posterior pole or midperiphery. Retinal hamartomamay progress from a flat lesion to a more elevated lesion without symptomatic changes in approximately 9.7%. Visual loss is more likely to result from optic nerve damage secondary to chronic papilledema caused by increased intracranial pressure (ICP). The increased ICP is caused by obstruction of the foramina of Monro by subependymal giant-cell astrocytoma. The frequency of this phenomenon in TSC is believed to be under 5 %, but its effects on vision may be devastating.

CONCLUSION

Astrocytomas in the macula can cause amblyopia. Subependymal nodules and cortical tubers cause hydrocephalus and blockage of Foramen of Monro leading to raised intracranial pressure. These manifestations can cause irreversible visual loss due to optic nerve damage. Timely recognition of the signs of raised intracranial pressure and early reference to neurosurgeon help in preventing visual loss. Most patients are mentally challenged and unable to voice complaints of raised intracranial pressure such as headache, nausea and altered consciousness. Hence, periodic ophthalmic examination is necessary to prevent visual loss.

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