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Case Report

Case Report- A Case of Tuberous Sclerosis

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Abstract

Introduction: Tuberous sclerosis complex (TSC) is an autosomal dominant phakomatosis. Tuberous sclerosis is a disorder characterized by the growth of numerous benign tumours in many parts of the body caused by mutations of TSC1 and TSC2 genes. This rare genetic disorder is usually associated with a triad of seizures, mental retardation and cutaneous lesions [1].

Case Description: The case of a 11 year old male presenting with loss of consciousness 1 month back. On examination he was diagnosed to be a case of tuberous sclerosis. Over the skin of the face there was adenoma sebaceum. There was a hypo pigmented patch present over the left lumbar region, face, fore limb. There was a shagreen patch over the back. Fundus examination showed hamartomas with mild exudation in the right eye. This case report emphasizes the importance of complete evaluation of a case presenting with loss of consciousness. TSC should be considered in children presenting with seizures, developmental delay and mental retardation.

Conclusion: Adenoma sebaceum and retinal hamartomas are very common ocular manifestations of Tuberous Sclerosis. Majority of the retinal hamartomas are non-progressive but lesions with sub retinal fluid and progression to total exudative detachment are reported. So, all cases of tuberous sclerosis must be evaluated at an ophthalmic setup [2].

Keywords: Tuberous Sclerosis Complex (TSC); TSC1 and TSC2 Genes

Introduction

Tuberous sclerosis complex (TSC) is an autosomal dominant phakomatosis. Tuberous sclerosis is a disorder characterized by the growth of numerous benign tumours in many parts of the body caused by mutations of TSC1 and TSC2 genes.

Case Report

An 11 year old male child was admitted in our hospital with a history of loss of consciousness 1 month back and the child's father gives a history of multiple warty growths over the face, which first started over the left upper eyelid and then gradually progressed to involve the whole face. He was born of non-consanguineous marriage with an uneventful birth history. There was no significant family history. The patient was well nourished, conscious, cooperative and well oriented. The child had multiple hyper-pigmented papules over the face (adenoma sebaceum). He also had two hypopigmented macules (ash leaf) over the lumbar region along with a Shagreen patch over the back. On the CNS examination no abnormality was detected.

On ophthalmological examination visual acuity was 6/6 in both eyes. On slit-lamp examination anterior segment appeared normal and on dilated fundus examination there were hamartomas along the inferior arcade with mild exudation in the right eye and left eye appeared normal.

On MRI brain plain it showed multiple cortical and subcortical ill-defined hyperintensities and subependymal calcified nodules and subependymal hamartomas. Other investigations like haemoglobin, complete blood count, renal and liver function tests were normal.

Figure 1: Adenoma sebaceum ashleaf spots.

Figure 2: Shagreen patch retinal hamartoma.

Discussion

Tuberous sclerosis or tuberous sclerosis complex (TSC), one of the phakomatoses, is characterized by hamartomas of the heart, kidney, brain, skin and eyes. Tuberous sclerosis complex (TSC) has retinal and non-retinal ophthalmic manifestations [4]. Retinal hamartomas are common ophthalmic manifestation and are noted in 50 - 80% of patients. Shields., *et al.* presented four cases, in children between the ages of 1 and 14 years, with TSC with peripapillary astrocytic retinal hamartomas that presented an 'aggressive behaviour' characterised by persistent growth, development of exudative retinal detachment, neovascular glaucoma, ocular pain, absence of light perception and that ended in enucleation of the eyeball [3]. Diagnostic Criteria for TSC is based on major and minor criteria. Definite TSC can be made when two major or one major plus two minor features are demonstrated.

Major criteria	Minor criteria
1. Cortical tuber	1. Cerebral white matter migration lines
2. Subependymal nodule	2. Multiple dental pits
3. Facial angiofibroma or forehead plaque	3. Gingival fibromas
4. Ungual or periungual fibroma (nontraumatic)	4. Bone cysts
5. Hypomelanotic macules (>3)	5. Retinal achromatic patch
6. Shagreen patch	6. Confetti skin lesions
7. Multiple retinal hamartomas	7. Nonrenal hamartomas
8. Cardiac rhabdomyoma	8. Multiple renal cysts
9. Renal angiomyolipoma	9. Hamartomatous rectal polyps
10. Pulmonary lymphangioleiomyomatosis	

Table

Our patient had two major criteria (subependymal nodules in MRI brain, Shagreen patch) 2 minor criteria (confetti skin lesions, retinal achromatic patch) which fit in the diagnosis of Tuberous sclerosis [5].

Conclusion

Retinal hamartomas can present with different symptomatology and signs. Aggressive retinal hamartomas can progressively enlarge and result in total exudative retinal detachment and neovascular glaucoma [2]. A complete ophthalmic evaluation and subsequent follow up are key to successful management of Tuberous sclerosis presenting with ocular complaints.

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