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OCULAR FINDINGS OF TUBEROUS SCLEROSIS – A CASE REPORT

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ABSTRACT

The term “Neurocutaneous syndromes” also known as Phakomatoses refers to a broad category of congenital diseases that includes anomalies of neuroectodermal and occasionally mesodermal development, hence commonly involving the skin, eyes and central nervous system. The term Phacomatoses was coined by Van Der Hoeve in 1921. Tuberous sclerosis is one type of Neurocutaneous syndromes. The word "tuberous sclerosis" comes from the distinctive cortical tubers that Désiré-Magloire first described. Tuberous sclerosis is a hereditary disorder that affects numerous body systems. A 30 year old female diagnosed as Tuberous sclerosis was referred by Dermatology Department. Detailed ophthalmic examination was done. Patient was diagnosed with Adenoma sebaceum of upper eyelids in both eyes and multiple retinal hamartoma in left eye.

KEYWORDS: Tuberous sclerosis, Adenoma sebaceum, Hamartoma, Phakomatoses

INTRODUCTION

Tuberous Sclerosis Complex (TSC) is a multisystemic autosomal dominant disorder which is characterized by hamartomatous development in any organ such as skin, central nervous system, kidney and lung. It is also known as Bourneville Disease as was first described by Bourneville in 1880. In 1908 Vogt has established triad of Tuberous Sclerosis – this includes Epilepsy, mental retardation and a sebaceous adenoma. This disease is often not diagnosed because of their wide phenotypic variability. The diagnosis is made using both genetic and clinical criteria, including the detection of tumor suppressor gene TSC1 and TSC2 inactivating pathogenic mutations and also comprising the cutaneous, renal, pulmonary, cardiac and neurological system manifestations. Other characteristic features of tuberous sclerosis are Ash leaf sign, shagreen patches, cortical tubers, retinal astrocytoma, facial angiofibromas and renal angiomyolipomas. Other than adenoma sebaceum of the lids, the ocular symptoms of tuberous sclerosis include a range of nonretinal ophthalmic abnormalities. Retinal or optic nerve hamartomas are present in almost half of tuberous sclerosis patients, and they are bilateral in 50% of these patients. They are now called Astrocytic Hamartomas.

CASE HISTORY – A 30 year old female diagnosed as Tuberous Sclerosis was referred from Dermatology department. At the age of the 15years she had developed facial angiofibromas, periungal angiomas and had 2 episodes of seizures when she was 17 years old. She had positive family history of facial angiofibromas and confetti lesions in her mother.

During the general examination she was conscious, cooperative and well oriented to time, place and person. Her vitals was within the normal limits.

Systemic examination was done and revealed the presence of adenoma sebaceum, subungual fibroma and Confetti skin lesions over leg.

On Ophthalmic examination – her best corrected Snellen's visual acuity was 6/6 in both the eyes. Angiofibromas were present in both upper eyelids. Her intraocular pressure (IOP) on Applanation Tonometer was normal in both the eyes. Rest anterior segment of the both the eyes appear normal. Optic disc and retinal examination was done after dilating pupil of both the eyes. Right eye having normal optic disc and macula on fundus examination, while left eye shows multiple, small and smooth surface hamartoma present superior and inferior to the disc along the blood vessels and one hamartoma present just below the macula.

Optical Coherence Tomography (OCT) was done of this patient. OCT scan shows a hyper-reflective, homogenous, raised, fusiform lesion located on the superficial layers of retina without any underlying shadowing. On OCT, the choroid and outer layers of the retina were plainly discernible. These attributes were indicating a retinal hamartoma.

DISCUSSION:

Tuberous sclerosis or tuberous sclerosis complex (TSC) is rare, multi-system hereditary disease that causes Non-malignant tumors to develop in the brain and other important organs such as heart, lungs, eyes, kidney and skin. A variety of symptoms seizures, developmental delays, and behavioural issues, skin abnormalities, lung and kidney disease. A mutation in one of the two TSC1 and TSC2 genes, which respectively encode for the proteins tuberin and hamartin results in TSC. These proteins acts as tumor growth suppressors agents that regulate cell proliferation and differentiation. According to various studies, the tubers found in various organs in this complex exhibit significant angiogenic qualities and a high surface expression of vascular endothelial growth factor (VEGF), which may be the cause of the neovascularization that mimics proliferative retinopathy. Both retinal and non-retinal ocular manifestations can occur with Tuberous sclerosis complex. The Non-retinal abnormalities include angiofibromas of the lids, colobomas of the iris, lens and choroid, strabismus, poliosis of the lashes, papilloedema and sector iris depigmentation. Retinal Hamartoma (RH) which are present in 50-80% of individuals, are the most prevalent abnormality found amongst the ocular manifestations of the TSC. Retinal hamartomas could be of 3 types – 1) smooth, flat, grey, translucent, non-calcified. 2) Mulberry like, raised, multinodular, calcified, opaque. 3) A lesion in transition with the hallmarks of the first two.

The flat variety RHs have a pale or faint in color; these lesions could be modest, clinical examinations frequently miss them. TSC are aggressive hamartomas with gradual, stagnant growth is possible. Because of this reason periodic retinal imaging is required to recognise

these lesions, track their development and growth. The hamartomas may be highly vascularized. Although they do not enlarge, some lesions become calcified with the time. Retinal and optic nerve hamartomas rarely causes vision loss. Due to development and progression of fundus lesions are uncommon, therapy is not necessary.

Other retinal findings include hyper-pigmented regions (likely caused by congenital retinal pigment epithelium hypertrophy) and retinal pigmentary disruption to "punched out" regions of hypopigmentation at the mid periphery or posterior poles.

Some interventional studies for macular edema related vision loss have demonstrated sub retinal fluid absorption following intra-vitreal Anti VEGF injections, Argon laser photocoagulation and photodynamic therapy. Fortunately, our patient didn't have any neovascularization, macular edema or since there was no calcification and the vision was still intact, we recommended her routine follow up and imaging scan of the brain to look for any tuberous as she had past history of seizure.

It's also critical to understand that, despite the absence of a treatment for the illness, symptoms can be managed. Hence, awareness and education regarding different organ manifestations of Tuberous sclerosis is crucial in the management of the patient as a whole.

CONCLUSION:

Retinal hamartomas in majority of the cases are non-progressive but sometimes this lesion with sub retinal fluid and can progress to total exudative detachment are reported. Therefore detailed ocular examination is mandatory in each and every case of Tuberous sclerosis with timely follow up. It is advised to use a multidisciplinary strategy to improve clinical outcomes.

Table – 1 : Revised diagnostic criteria for Tuberous sclerosis complex

Major features	Minor features
Facial angiofibromas	Dental enamel pits
Periungual fibromas	Rectal polyps
Shagreen patch	Bone cysts
Retinal hamartomas	Gingival fibromas
Cortical tubers	Non-renal hamartomas
Subependymal nodule	Renal achromic patch
Cardiac rhabdomyoma	Confetti skin lesions
Renal angiomyolipoma	Renal cyst
	Cerebral cortical dysplasia

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CONFLICT OF INTEREST: NIL

REFERENCE:

1. Bourneville DM. Sclerose tubereuse der circonvolutions cerebrales: idiotie et epilepsie hemiplegique. Arch Neurol (paris) 1880;1:81-91

2. IJSR, ocular manifestations in Tuberous sclerosis-A case report, vol- 8,issue-5,May-2019,Page-64.

3. IJO, primary ocular presentation of Tuberous sclerosis- A case report, Nancy M Rajasekaran, subam horo, Thomas kuriakose, March 2019, pp433.

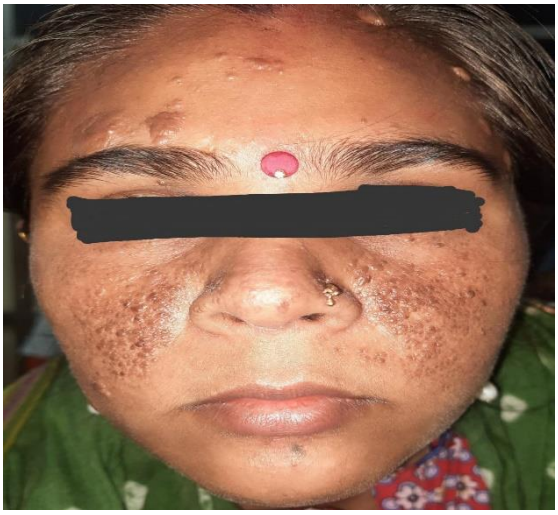

Figure 1 Adenoma Sebaceum

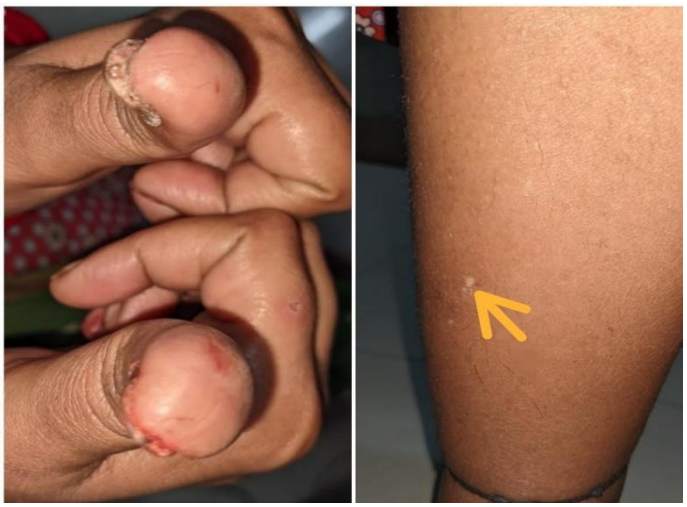

Figure 2 Periungual fibroma and Confetti lesions

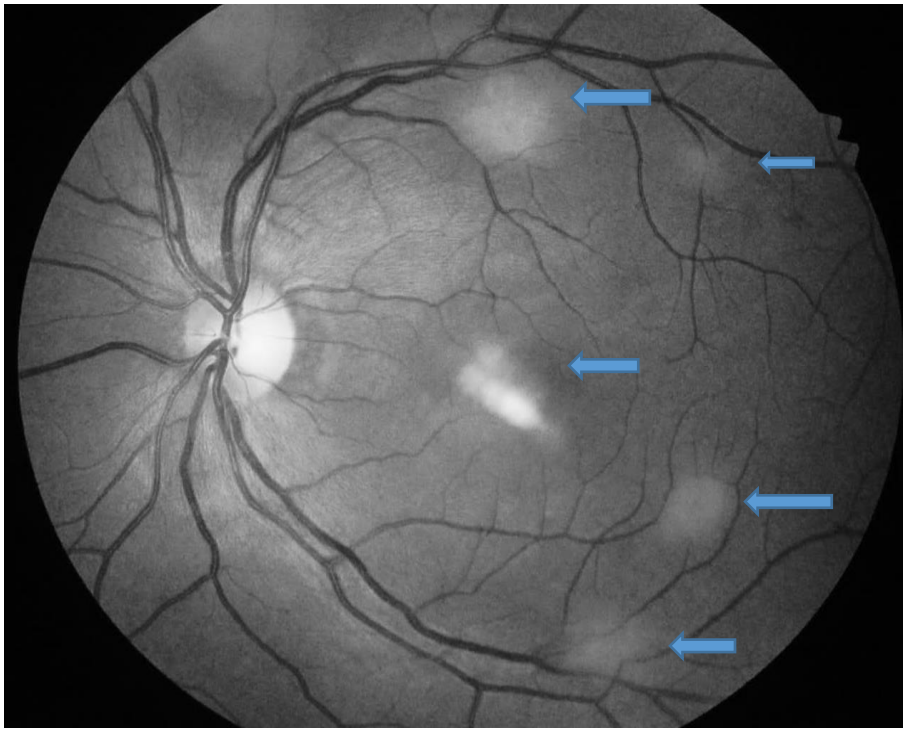

Figure 3 Left eye shows multiple, small and smooth surface hamartoma present.

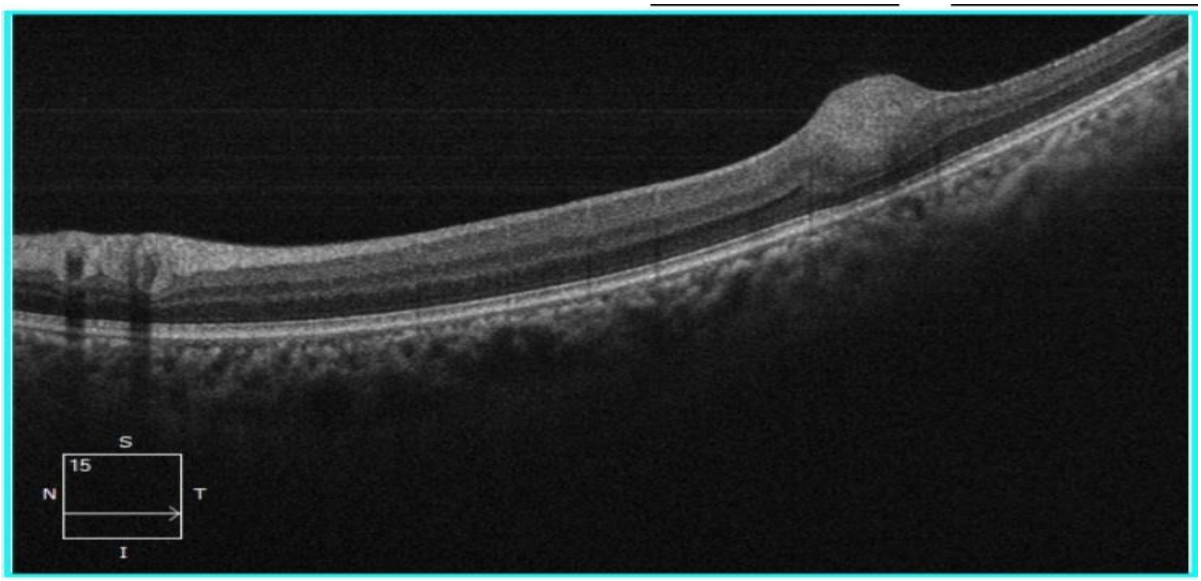

Figure 4 The Optical coherence tomography (OCT) Scan was done, shows Hyper-reflective, homogenous, elevated, fusiform lesion located on the Superficial portion of the retinal layers with no shadowing, outer retinal layers and choroid clearly seen on the OCT scan. These features are suggestive of Retinal Hamartoma.