



OCULAR MANIFESTATIONS IN TUBEROUS SCLEROSIS- A CASE REPORT

Ophthalmology

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ABSTRACT

Tuberous Sclerosis is a multisystem disorder characterised by hamartomatous growth in any organ. Ophthalmic manifestation can be retinal or non-retinal. A 24 yr. old female diagnosed as Tuberous sclerosis was referred from Dermatology department. Full Ophthalmic examination was done. Patient was diagnosed with adenoma sebaceum of lid and retinal hamartoma in right eye.

KEYWORDS

Tuberous sclerosis, Hamartomas, Smooth retinal hamartomas

CASE REPORT

INTRODUCTION– Tuberous Sclerosis Complex (TSC) is a multisystem disorder characterised by hamartomatous growth in any organ. It has autosomal dominant inheritance. It is also called Bourneville Disease as was first described by Bourneville in 1880. Vogt in 1908 described the classic triad- epilepsy, mental retardation, adenoma sebaceum. Other characteristic features are Ash leaf sign, shagreen patches, cortical tubers, retinal astrocytomas, facial angiofibromas and renal angiomyolipomas. In 1921 Van der hoeve termed the retinal lesions as phakomas (spots), they are now called astrocytic hamartomas.

CASE HISTORY – A 24 year old female diagnosed as Tuberous sclerosis was referred from dermatology dept. She had facial angiofibromas since 15 years, periungual angiomas since 13 yrs, seizures since 9 yrs and gingival fibroma since 3 yrs. She had family history of fibroma in younger sister. Positive Personal history of poor performance in school was present.

On General examination she was conscious, co-operative and well oriented to time, person and place. Her vitals were normal.

On Ophthalmic examination- her best corrected Snellen's visual acuity was 6/6 in both eyes. Angiofibromas were present in both lower lids. Rest anterior segment and vitreous cavity were normal.

On fundus examination (after full dilatation with tropamide 1% and phenylephrine 5%) optic disc and retinal examination in left eye were normal. In right eye fundus examination demonstrated a flat, smooth hamartoma inferior to disc.

Systemic examination revealed the presence of adenoma sebaceum, gingival fibroma, subungual fibroma.



Fig.1: Adenoma Sebaceum

Fig.2 Periungual Fibroma



RIGHT EYE

DISCUSSION –

In TSC Ophthalmic manifestation can be retinal and non-retinal. The non-retinal findings are angiofibromas of lids, coloboma (of iris, lens, choroid), strabismus, poliosis of lashes, papilloedema, sector iris depigmentation.

Retinal hamartomas may be of 3 types- a) flat, smooth, non-calcified, grey, translucent. b) elevated, multinodular, calcified, opaque, mulberry-like. c) transitional lesion with features of above two.

Refraction – visual loss is rare, but seen if ->

- Large retinal hamartomas.
- Optic nerve involvement.
- Neovascularisation of retina with macular edema.
- Intracranial tumours.
- Intracranial hypertension.

Rarely sub-retinal fluid accumulation progressing to total exudative retinal detachment and calcification has been documented over time.

Investigations– fundus exam/ dermatology exam/ CT/ MRI/ examination of family members.

Symptomatic treatment is required till vision is not affected and strict follow-up should be advised.

For deterioration of vision with macular edema, some interventional studies have shown resorption of subretinal fluid after Argon laser photo co-agulation, intravitreal anti-VEGF, photodynamic therapy. Our patient fortunately had no macular edema, neovascularisation or calcification and the vision was well preserved hence we advised her regular follow up.

DIFFERENTIAL DIAGNOSIS- can be neurofibromatosis, retinoblastoma, choroidal osteoma, choroidal metastasis, coats disease.

CONCLUSION – Adenoma sebaceum lid and retinal hamartomas are the most 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 ophthalmic examination of all cases of Tuberous Sclerosis is mandatory with a regular follow-up.

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