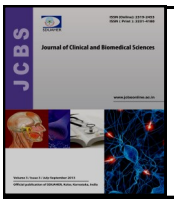

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Case Report

Tuberous Sclerosis: Is Cataract a New Association?

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Abstract

An eight year old female child was admitted with history of four episodes of partial seizures since one day, reddish brown papular rashes over malar region of face and chin in a butterfly pattern suggestive of adenoma sebaceum, hypo pigmented macules on the trunk of ash leaf spots type and diminution of vision in left eye since 5 years. On slit lamp examination of the left eye an iris coloboma and cataractous lens was found. Fundus examination of the right eye showed an edematous disc and a retinal astrocytoma in the superonasal quadrant. A diagnosis of tuberous sclerosis (TS) was made. Retinal and optic nerve involvement in TS are well known today with approximately 50% manifesting with retinal hamartomas. Unusual findings reported are band keratopathy, keratoconus and cataract. A review of the literature indicates that although in some cases, cataract is attributable to local ocular disease, or congenital causes, it may be a rare manifestation of TS.

Key words: tuberous sclerosis, cataract

Introduction

Tuberous sclerosis complex (TSC) is a multisystem disorder characterised by hamartomatous growths that can occur in almost any organ or tissue. [1] The first description of TSC is usually attributed to Bourneville in 1801 and in 1908, Vogt described the classic triad of epilepsy, mental retardation and angiofibroma earlier called as adenoma sebaceum. The clinical manifestations of TSC are now known to be far more diverse These include cortical tubers, retinal astrocytomas, facial angiofibromas, ungual fibromas, hypomelanotic skin lesions, shagreen patch and renal angiomyolipomas. Ophthalmic features associated with TSC can be divided into retinal and non-retinal. The retinal associations of TSC pharomatosis which are now known as astrocytic haemartomas were first noted by Van der Hoeve in 1921. Three basic morphological types of retinal hamartomas are described in the literature. (i) the relatively flat, smooth, non-calcified, grey, hyperpigmented areas (probably congenital retinal pigment epithelium hypertrophy) to "punched out" hypopigmented areas at the posterior pole or mid of periphery. Other findings include angiofibromas of the eyelids, coloboma of the iris, lens and choroid, strabismus, poliosis of eyelashes, papilloedema and sector iris depigmentation. In addition to retinal lesions in patients with tuberous sclerosis, unusual findings such as band keratopathy and keratoconus have been reported. Cataracts associated with tuberous sclerosis have been described in very few case series.

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Case History

An eight year old female child was admitted in Paediatrics ward with history of four episodes of partial seizures since one day, multiple blackish raised lesions over the face since two months and diminution of vision in left eye since five years. There was no history of trauma to the eye. Child was delivered at home by a full term vaginal delivery and had cried immediately after birth. There was no history of maternal fever or drug intake during pregnancy. Developmental history was normal for her age. There was no history of similar complaints in family members. On general physical examination, the eight year old girl was moderately built and nourished for her age, was conscious, cooperative and well oriented to time and place. The vital parameters and clinical systemic examination

were within normal limits. Reddish brown papular rashes over the malar region of face and chin in a butterfly pattern were found which is typical of adenoma sebaceum (Fig 1). Hypo pigmented macules on the trunk of tyre ash leaf spots type were observed. Visual acuity of the right eye (RE) was 6/6 and in the left eye (LE) was able to count the fingers at 2 metres. On slit lamp examination of anterior segment of the LE an iris coloboma and a cataractous lens was found (Fig 2).

Fundus of the RE showed an oedematous disc and a well defined, solitary hypopigmented lesion in superonasal quadrant away from the disc suggestive of an astrocytic retinal hamartoma (Fig 3). B scan showed echogenous cataractous lens. A non-contrast CT of the brain showed calcification of sub ependymal nodules and cortical tubers (Fig 4). An IQ score of 75 was elicited on applying inet Kamat Scale which is a borderline score.

Based on the current Tuberous Sclerosis Alliance criteria (Table1) a diagnosis of Tuberous Sclerosis was made. The cataractous lens was extracted under general anaesthesia after physical clearance obtained from the paediatrician and a secondary intra ocular lens implantation was to be planned at a later date. On follow up of the girl after six months the vision in LE was 6/60 and in the RE the retinal hamartoma remained of the same size.

Fig1. Adenoma Sebaceum over the face

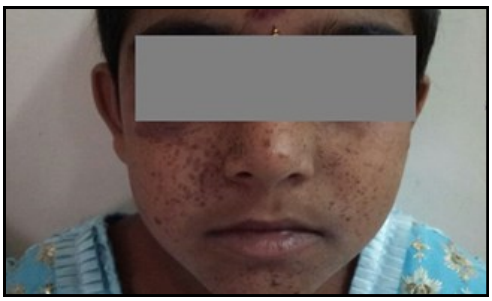

Fig 2. Cataract in the left eye

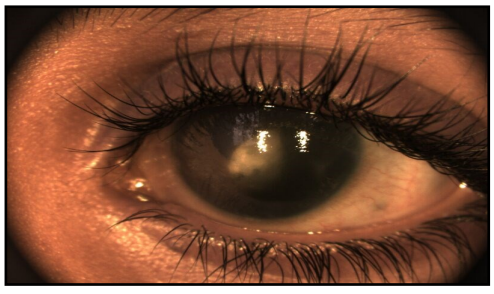

Fig 3. Astrocytic retinal hamartoma of the right eye

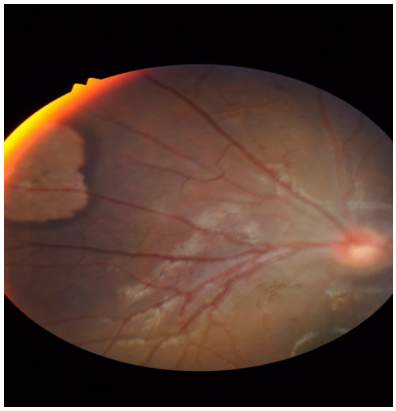

Fig 4. C T Brain showing calcificatian of sub ependymal nodules and cortical tubers.

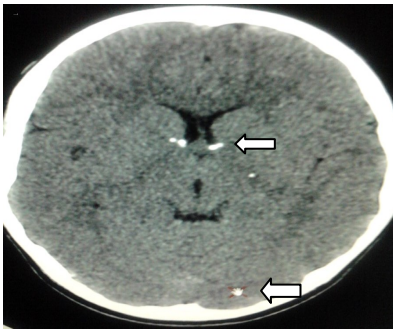

Table 1. Tuberous sclerosis alliance criteria

Major features
Facial angiofibromas
Hypomelanotic macules
Shagreen patches
Cortical tubers
Subependymal nodules
Subependymal giant cell tumours
Retinal hamartomas
Cardiac rhabdomyomas
Renal AML
Pulmonary LAM
Minor features
“confetti” skin lesions
Gingival fibromas
Pits in dental enamel
camclines
Retinal achromatic patches
Bone cysts
Hamartomatous rectal polyps

Table 2. Non-retinal ophthalmic lesions reported with TS

Ocular tissue	Lesions
Eyelid	Angiofibroma, vitiligo, poliosis, ptosis, epicanthic fold
Conjunctiva	Grey with flat surfaced pedunculated tumour
Cornea	Megalocornea, keratoconus, band keratopathy, pannus, posterior embryotoxin
Anterior chamber angle	Glaucoma
Iris	Rubeosis, iridocyclitis, coloboma, sector depigmentation
Ciliary body	Neoplasm
Choroid	Coloboma, angioma
Lens	Subluxation**,** cataract, coloboma
Pupil	III nerve paresis
Vitreous	Hamartomatous fragments, haemorrhage, pseudoglioma
Globe	Enopthalmos, myopia, phthisis
Extraocular muscles	Non paralytic strabismus, paralysis of cranial nerves III & VI, nystagmus

Discussion

Eye manifestations are common in TS and 50% have retinal hamartomas.[2] Foci of fundus and iris depigmentaion and pedunculated conjunctival lesions have also been described. Papilloedema may be seen as expanding brain tumours may elevate the intra cranial pressure. Additional unusual findings, such as band keratopathy and keratoconus have been reported. There have been only a few reported instances of cataract in TS. A study done by Acher and Nevin reported on the non-retinal ophthalmic manifestations of TS (Table 2).[5] Only one of Robertson's patients had cataract, and no details of the case are available (personal communication May 1995).[6] Three reports of single cases of TS note band keratopathy and cataract. In these cases, the cataract might have been secondary to some other local eye disease and not a primary manifestation of TS. Yakovlev described a case of bilateral congenital cataracts in the disease. In 1940 and 1946, Hall encountered a 17 year old boy with TS who had peripheral degeneration of both lenses and a small nodular mass in the centre of one lens. These abnormalities did not change during 4.5 years of observation. The lesion could have been a congenital cataract. Ross and Dickerson described a unilateral posterior polar cataract in a 19 year old with TS and bilateral keratoconus. Walsh observed opaque nodules in the subcapsular cortex in a 16 year old boy with TS. In 1996, two cases of TS with unilateral cataracts were reported by Judith E.A.Warner and Simmons Lessell.[2] Bilateral punctate cataracts were reported by Sunil Ganekal, Syril Drairaj in 2014.[4]

Although the occurrence of cataract may be coincident, we recommend that young patients with TS be screened for lenticular opacities and followed up to prevent the development of visually disabling cataracts.

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