A rare case of visual loss due to serous detachment associated with retinal “mulberry” hamartoma in a case of tuberous sclerosis

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Introduction

Tuberous sclerosis or tuberous sclerosis complex (TSC) is a rare multisystem genetic disease that causes nonmalignant tumors to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs, and skin. A combination of symptoms may include seizures, developmental delay, behavioral problems, skin abnormalities, lung, and kidney disease. Ocular involvement commonly occurs in tuberous sclerosis though visual loss is rare. Retinal lesions called astrocytic hamartomas (or “phakomas”), appear as a smooth surface or nodular grayish or yellowish–white lesion in the back of the globe on the ophthalmic examination. Astrocytic hamartomas can calcify. Nonretinal lesions associated with TSC include coloboma, angiofibromas of the eyelids, and papilledema.

Case history

A 7-year-old male child presented at our outpatient department with gradual visual loss since 2 years in his right eye. The child showed delayed developmental milestone. The parents gave a history of similar affected sibling in the family. Dermatological examination revealed reddish spots or bumps, on the nose and cheeks

in a butterfly distribution (facial angiofibromas or “adenoma sebaceum”) and hypomelanotic macules (“ash leaf spots”) or patchy depigmented areas in different parts of the body (Fig. 1). An area of thick leathery skin that dimpled like an orange peel was found on the lower back (Shagreen patch; Fig. 2). Detailed ophthalmological examination was done. Visual acuity was reduced in right eye 20/200 but it was 20/20 in left eye. Slit lamp examination of anterior segment of both eyes was normal. Dilated fundus examination was done by +90 dioptre slit lamp biomicroscopy. Right eye fundus showed presence of a grayish yellow mulberry like mass adjacent to the optic disk at its superior pole (Fig. 3). The foveal reflex in the right eye was dull with circumscribed round elevation at the macula. The left eye fundus was normal. Optical coherence tomography showed serous detachment of sensory retina from the retinal pigment epithelium at the subfoveal region in right eye while of left eye it was normal (Fig. 4). The child declined to undergo digital fluorescein angiography. Echocardiography and renal ultrasound were normal. Magnetic resonance imaging of brain was also normal. A diagnosis of tuberous sclerosis with associated retinal hamartoma and serous detachment was made based on the positive family history, classic dermatological findings, and fundus findings. Visual loss was attributed to the retinal hamartoma associated with subfoveal serous detachment in right eye.

Discussion

TSC is a multisystem disorder characterized by hamartomatous growths that can occur in almost any organ or tissue.

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Fig. 1 Photograph showing facial angiofibromas or adenoma sebaceum

Fig. 3 Fundus photograph showing retinal hamartoma or mulberry tumor at the superior pole of optic nerve head

The first description of TSC is usually attributed to Bourneville in 1880 and in 1908, Vogt described the classic triad of epilepsy, mental retardation, and adenoma sebaceum (now called angiofibromatosis) [1, 2]. The clinical manifestations of TSC are now known to be far more diverse and a more inclusive system of primary and secondary diagnostic criteria has been introduced. These include cortical tubers, retinal astrocytomas, facial angiofibromas, ungual fibromas, hypomelanotic skin lesions, shagreen patch, and renal angiomyolipomas [3].

Ophthalmic features associated with TSC can be divided into retinal and non-retinal. The retinal associations of TSC were first noted by Van der Hoeve in 1921 [4]. He termed these lesions phakomas (derived from the greek *phakos*, meaning spot) and introduced the concept of phakomatosis. These retinal lesions are now known to be astrocytic hamartomas [5, 6]. Three basic morphological types of retinal hamartomas are described in the literature: (1) the

relatively flat, smooth, noncalcified, gray, translucent lesion; (2) the elevated, multinodular, calcified, opaque lesion resembling mulberries; and (3) a transitional lesion which has morphological features of both of the previous. Other retinal findings include retinal pigmentary disturbance ranging from hyperpigmented areas (probably congenital retinal pigment epithelium hypertrophy) to “punched out” hypopigmented areas at the posterior pole or midperiphery [7–9]. Nonretinal findings include angiofibromas of the eyelids, coloboma of the iris, lens and choroid, strabismus, poliosis of eyelashes, papilloedema, and sector iris depigmentation. Visual loss is rare in tuberous sclerosis but may occur from retinal or optic nerve involvement or from intracranial tumors affecting the part of the brain that processes the visual information. Visual decline may also accompany subependymal giant cell astrocytomas of the optic nerve. In this case, the child developed visual loss in his right eye due to subfoveal serous detachment along with astrocytoma adjacent to optic nerve head.

Fig. 2 Photograph showing Shagreen patch of the lower trunk

Fig. 4 Optical coherence tomography of right eye showing serous detachment

Visual loss due to serous retinal detachment in patients of tuberous sclerosis alone has rarely been reported in literature [10].

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