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Photo Essay

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Case Report

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Abstract

A case of tuberous sclerosis with B/L Iris coloboma, Right Eye Total cataract with Sensory exotropia. Left Eye Astrocytic hamartoma with Adenoma sebaceum and Shagreen patch as systemic associations.

Keywords: Tuberous Sclerosis; Cataract; Adenoma Sebaceum; Iris Coloboma

Introduction

A 16-year-old boy presented to our OPD with complaint of decreased vision in right eye since early childhood. On examination, visual acuity in R/E was HMCF, PR accurate and in L/E was 20/200. Slit lamp examination showed iris coloboma with total cataractous lens in R/E (Figure 1) while L/E had iris coloboma only (Figure 2). Fundus was not visible in R/E but L/E fundus showed retinal astrocytic hamartoma superonasal to disc. There was R/E exotropia of 30^0. Systemic examination revealed adenoma sebaceum over the face and shagreen patch over the back. These findings confirmed the diagnosis of tuberous sclerosis. Anterior segment findings like iris coloboma with cataract and non paralytic strabismus are not common presentations in patient with tuberous sclerosis in addition Astrocytic Hamartoma in this patient makes this case unique.

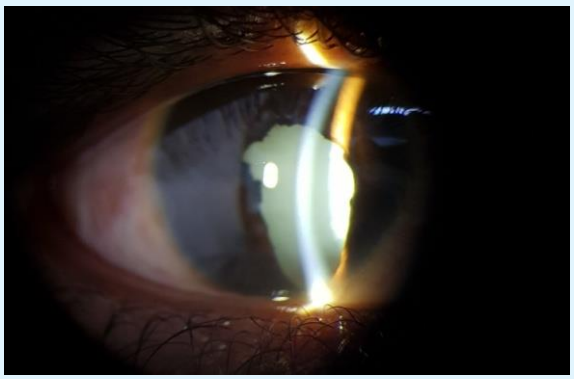

Figure 1: Iris Coloboma with Cataractous Lens.

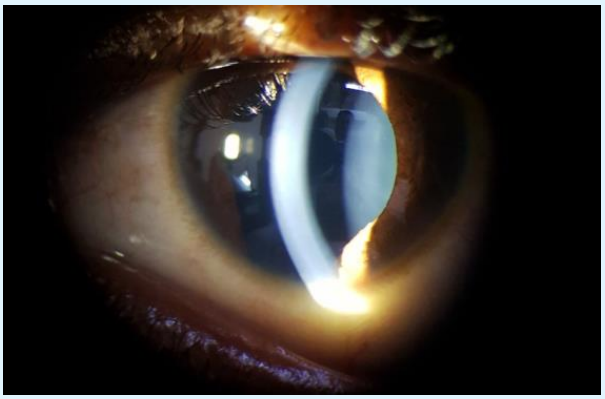

Figure 2: Iris Coloboma.

Figure 3: Adenoma Sebaceum.

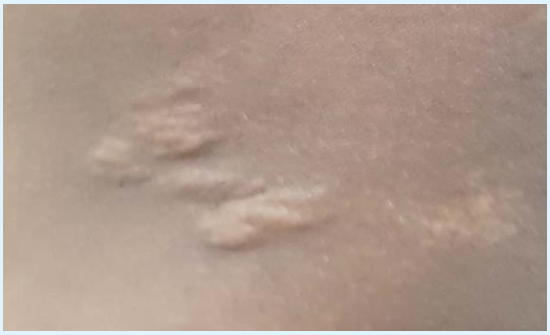

Figure 4: Shagreen Patch.

Discussion

Tuberous sclerosis complex (TSC) is a hamartomatous multisystem disorder involving nearly every organ of the body but primarily involving eye,skin and brain. TSC was initially described by Bourneville in 1880 and in 1908, the classic triad of epilepsy, mental retardation, and adenoma sebaceum was detailed by VOGT [1,2]. Clinical manifestations show a vast diversity. Major and minor diagnostic criteria for TSC have been established by the Tuberous Sclerosis Consensus Conference in 1998 (Table 1). Diagnosis of tuberous sclerosis is categorized according to presence of major and minor criteria (Table 2) [3].

Major Criteria	Minor Criteria
Angiofibromas (3 or more) or forehead plaque	Dental enamel pits (more than 3)
Hypomelanotic macules (3 or more)	Intraoral fibromas (2 or more)
Ungual fibromas (2 or more)	Nonrenal hamartomas
Shagreen patch or multiple collagenomas	Retinal achromic patch
Multiple retinal hamartomas	“Confetti” skin lesions
Cortical dysplasia’s (more than 3). This includes tubers and cerebral white matter radial migration lines.	Multiple renal cysts
Subependymal nodule(s)	
Subependymal giant cell astrocytoma(s)	
Cardiac rhabdomyoma	
Lymphangioleiomyomatosis (LAM)	
Angiomyolipomas (2 or more)	

Table 1: Diagnostic Criteria of Tuberous Sclerosis.

Definite TSC	2 major criteria or 1 major plus 2 minor criteria
Probable TSC	1 major and 1 minor criteria
Possible TSC	Either 1 major criteria or 2 or more minor criteria’s

Table 2: Major and Minor Criteria.

Ophthalmic manifestations associated with TSC can be categorized into retinal and non-retinal. The retinal lesions of TSC were initially termed as ‘*phakomas*’ by Van der Hoeve in 1921 and concept of phakomatosis was introduced following this. These retinal lesions are now known as *astrocytic hamartomas* which is the most common retinal finding [4,5]. Non-retinal findings include strabismus, poliosis of eyelashes, angiofibromas of the eyelids, coloboma of the iris, lens and choroid, papilloedema, and sector iris depigmentation [6,7]. The prevalence of TSC associated retinal astrocytic hamartomas varies though largest reported series from the Mayo Clinic gives an overall prevalence of 49% [8,9].

Prevalence of the non-retinal findings has been reported in a very few case series. Angiofibromas of the eyelids were the most common non-retinal finding (39%). Prevalence of non-paralytic strabismus and coloboma were only 5% and 3% [10]. Thus the presence of these very rare non-retinal findings like non- paralytic squint, B/L iris coloboma and cataract make our case exclusive.

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