

CASE REPORT**MACULAR COLOBOMA – IN A CHILD WITH USHER SYNDROME****Mazhar Ishaq, Ahsan Mukhtar, Saim Khan**

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Macular coloboma is a rare entity and its concomitance with Usher syndrome is described here. A 14 years male child was studied in detail along with other family members. He underwent two complete ophthalmologic examinations (4-years follow-up), including visual assessment, orthoptic evaluation, colour vision test, visual fields, corneal topography, Optical coherence tomography, fluorescein angiography, and electroretinography. Detailed ophthalmic examination was also conducted on other asymptomatic members of the same family. Patient had sensorineural deafness, poor visual acuity, and progressive visual field impairment in both eyes, bilaterally presenting macular coloboma and atypical retinitis pigmentosa pattern. The other investigated relatives did not show any specific and/or significant ocular disorder. This concurrence represents no genetic pattern and is observed in sporadic cases.

Keywords: Retinitis pigmentosa, Usher syndrome, Macular coloboma

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INTRODUCTION

Retinitis pigmentosa (RP) is a diverse group of inherited retinal dystrophies characterized by night blindness, visual field loss and retinal pigment deposits visible on fundus examination. Typical RP is characterized by the triad: arteriolar attenuation, waxy pallor disc, and bony spicule pigmentation.^{1,2} RP is rarely associated with various systemic syndromes. Usher syndrome which is characterized by sensorineural deafness is a distressing systemic association of RP. It is responsible for more than half of all the cases of combined deafness and blindness.^{2,3} Macular coloboma is a rare entity characterized by an excavated area of chorioretinal atrophy about 2–3 disc diameter in size.⁴ The proposed mechanism of its formation is thought to be the maldevelopment or an intrauterine infection.^{5–7}

This short communication presents a case of a child who presented with decreased vision and hearing loss. When examined in detail, it came out to be a case of Usher syndrome with a unilateral macular coloboma. We are unaware of any previous reports revealing association of macular coloboma with Usher syndrome and could find no reference to it in a computerized search utilizing MEDLINE.

CASE REPORT

A 14 years old boy who was hard at hearing presented with decreased vision in both the eyes (Left more than Right). Patient had been using glasses for the last 06 years but there was no birth history of antenatal infections, premature birth, any other systemic complaints, involvement of other family members with the same illness, or use of any drugs.

General physical examination revealed short stature, thin lean boy who was alert, active, well oriented in time, place and person, fully cooperative and conscious despite the fact that he was unable to hear or speak. Systemic examination of patient did not reveal

any abnormality. Ocular examination revealed best corrected visual acuity of 20/30 (6/9) in right eye and 20/1200 (1/60) in the left. Orthoptic examination revealed Lt exotropia of 15° with poor fixation of left eye. (Figure-2 and 3) Ocular movements were of full range and there was no nystagmus. Slit lamp examination revealed unremarkable anterior segments. Posterior segment revealed mid peripheral pigmentary changes, waxy disc in both the eyes and a well circumscribed, 4–5 disc diameter excavated area with overlying retinal vessels at the macula.(Figure-4 and 5) Optical Coherence Tomography (OCT) of Right eye showed decreased retinal thickness and macular atrophic changes.(Figure-6) OCT of left eye revealed a well circumscribed excavation and thinning of retinal layers. (Figure-7) Fundus Fluorescein angiography (FFA) of both the eyes showed hypo and hyper fluorescent areas all over the fundus secondary to pigmentary and atrophic RPE changes respectively, however FFA of Left eye also revealed a well circumscribed hypofluorescent area of about 4–5 disc diameter with absence of choroidal vasculature and choriocapillaris, typical for Macular coloboma.(Figure-8 and 9). Ultrasound B scan and A scan of left eye revealed a well circumscribed excavated area at macula (Figure-10), over which the axial length is increased to more than 24mm where normal length at the flat retina is 21 mm.(Figure-11) Brainstem Evoked Response Audiometry (BERA) report revealed profound hearing loss. All these clinical findings and the investigations confirmed the diagnosis of Left Macular coloboma in a patient with Type 2 Usher syndrome. Patient was counselled regarding the nature and prognosis of disease, advised glasses for refractive error, and referred to audiologist for hearing aids.

Examination of other family members, i.e., parents; elder sister and younger brother was unremarkable.

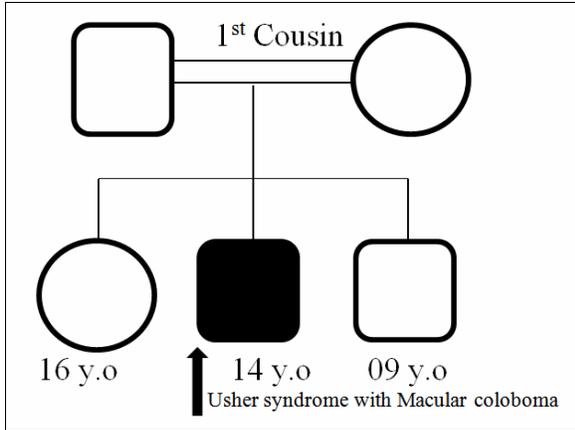


Figure-1: Pedigree of macular coloboma



Figure-2&3: 15° Exodeviation Lt, Poor Fixation Lt



Figure-4&5: (Right Fundus), (Left Fundus)

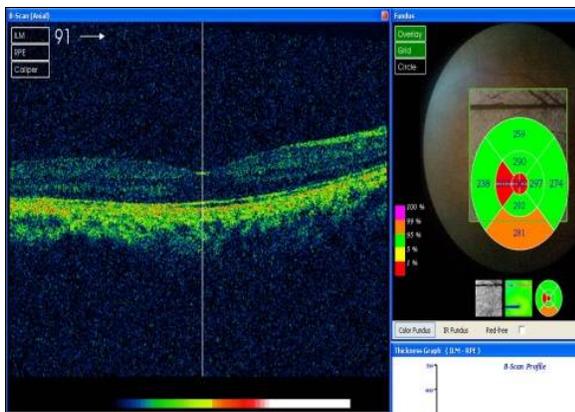


Figure-6: OCT right eye

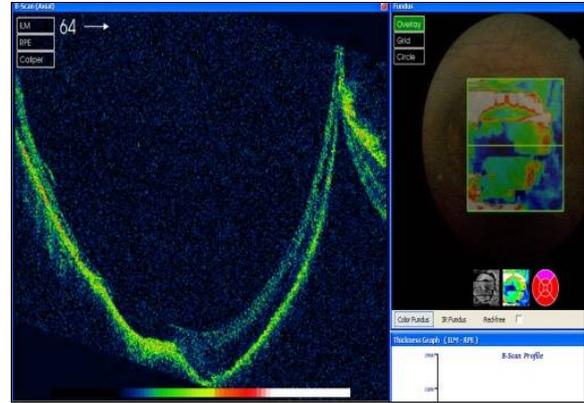


Figure-7: OCT left eye

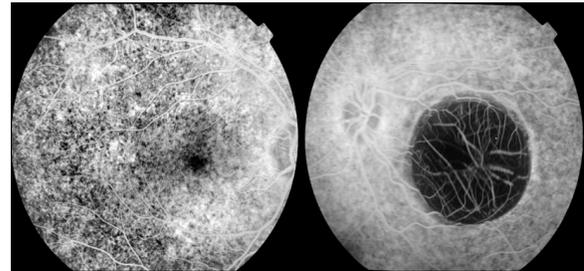


Figure-8&9: FFA right Eye, FFA left eye

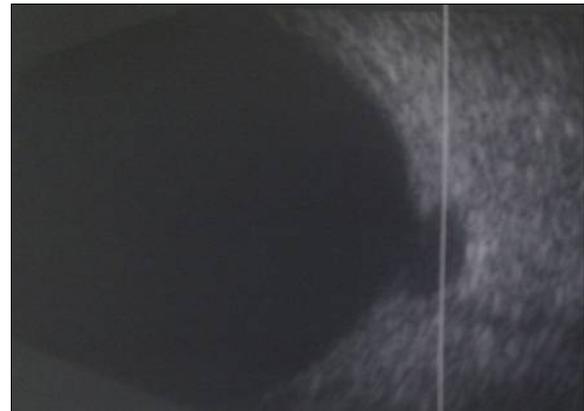


Figure-10: B-scan left eye–Arrow showing macular coloboma



Figure-11: A-scan of left eye- Arrow showing increased axial length

DISCUSSION

Macular coloboma is characterised by a well demarcated, large defect in the macular region oval or round in shape, and coarsely pigmented.^{6,7} Few reports in literature has demonstrated association of peripheral retinal changes such as Leber's congenital amaurosis, retinal dystrophy, retinitis pigmentosa (RP), or pigmented paravenous retinochoroidal atrophy with macular coloboma.⁸⁻¹⁰ Macular coloboma is classified into three types, namely pigmented macular coloboma, non-pigmented macular coloboma, and macular coloboma associated with abnormal vessels.¹¹ In our case it was a pigmented unilateral macular coloboma.

Usher Syndrome (USH) is an autosomal recessive condition and most common cause of combined deafness and blindness in children.^{2,3} Incidence of USH is 3 per 100,00 population and is characterized by sensorineural deafness and RP.^{12,13} It is important to attune the ophthalmologists to patients with RP who present with a nasal intonation to their speech or wear hearing aids and they must inquire when exactly the vision and hearing loss began. However, the hearing level of most Usher patients is stable over time.^{2,3} USH is classified into three types (Type 1, Type 2, Type 3) on the basis of age of onset and involvement of vestibular system. Type 1 is the most common one and accounts for 75% of the cases.¹⁴ Our patient had Type II USH as there was no history of vestibular dysfunction.

Association of Macular coloboma though documented with other hereditary disorders in literature but to best of our knowledge, this is the first reported case of Usher syndrome occurring along unilateral macular coloboma. Therefore, it is important for ophthalmologists to recognize Macular coloboma and differentiate it from mimicking lesions like toxoplasmosis scar, Macular cyst etc. It should be kept in mind that although rare Macular coloboma may be found along with USH and responsible for profound visual loss, this may influence the follow up and management.

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