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A Rare Case of Congenital Bilateral Macular Coloboma

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ABSTRACT
Coloboma of macula is a rare, non-syndromic developmental defect of the eye characterized by well-circumscribed, oval or rounded, usually unilateral, atrophic lesions of varying size presenting rudimentary or absent retina, choroid and sclera located at the macula leading to decreased vision and on occasion, other symptoms (e.g. strabismus). It is usually isolated, but may also be associated with Down syndrome, skeletal or renal disorders.This is a case of a 25-year-old male patient who presented to the outpatient clinic with a history of poor vision in both the eyes since childhood.

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I. INTRODUCTION

Coloboma is an ocular congenital defect that can affect different areas of the eye such as the iris, the retina, the choroid, the lens or the optic nerve. Macular coloboma is an atypical coloboma that is hypothesized to be the result of incomplete differentiation of the arcuate bundle along the horizontal raphe during development. Macular coloboma is a rare eye condition that may affect about 0.5-0.7/10,000 live births. On fundus examination, macular coloboma appears as well-demarcated atrophic lesions which can be unilaterally or in rare cases bilaterally. Patients affected with macular coloboma suffer from non-progressive decrease in visual acuity .Few cases of macular coloboma are associated with an inheritance pattern. The most common genetic inheritance is autosomal dominant while the rest of the cases are presented as an isolated cases with no known inheritance pattern. The condition may be associated with various syndromes such as patau syndrome, Fraser syndrome, franceschetti syndrome, cat eye syndrome and many others.

The purpose of this paper is to present the rare case of isolated bilateral congenital macular coloboma.

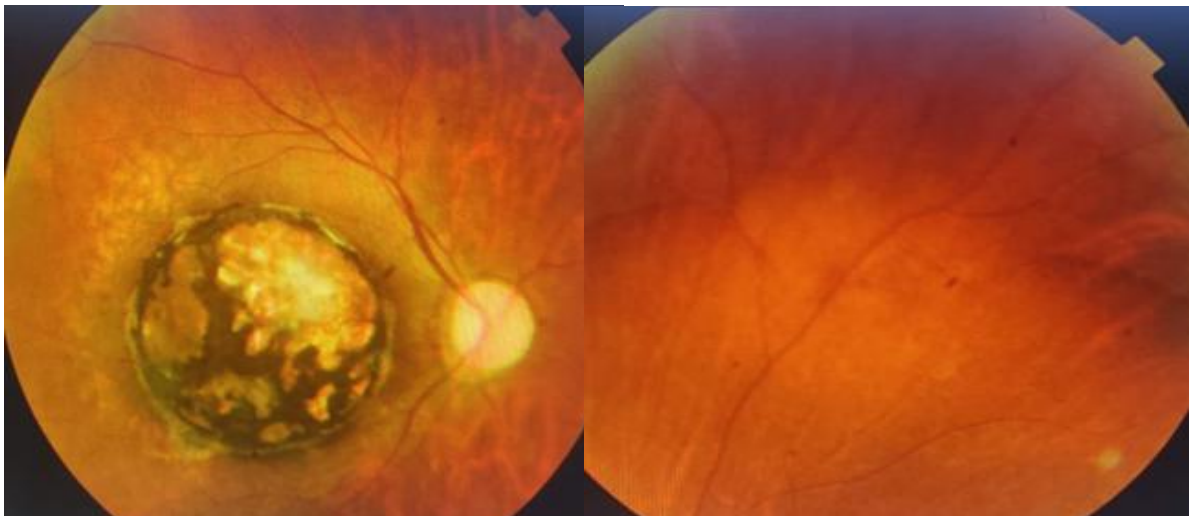
II. CASE REPORT

A 25-year-old male presented to the OPD with blurred vision in both eyes from childhood.There was no significant medical or family history, and it is non-consanguineous marriage for his parents. No syndromes or associations were detected. No history of any intrauterine infections.

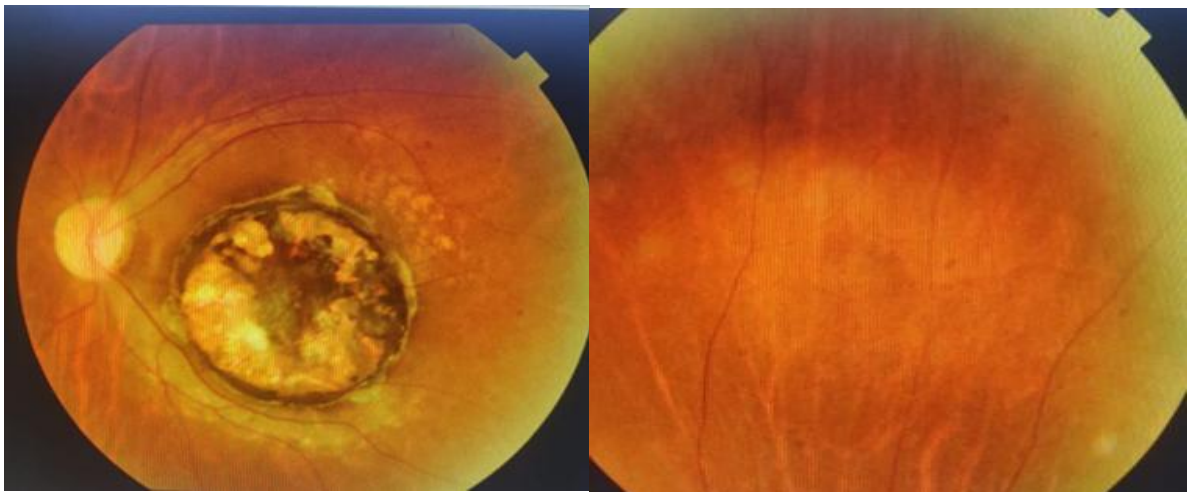
On Ophthalmic examination-Visual acuity in both eyes -1/60 and best corrected visual acuity is 3/60 with -10DS.Color vison-He was able to differentiate only primary colors. The Anterior segment Examination was unremarkable in both the eyes. Intra ocular Pressure (IOP) by applanation Tonometry was 18mmHg in both the eyes.Eye alignment by Hirschberg test and cover-uncover test showed orthotropic alignment in both eyes.Extra ocular movements were full,free,painless in all the gazes bilaterally. Axial length in right eye(OD)-26.74mm & left eye(OS)-27.00mm.

Fundus examination,using Indirect Opthalmoscopeshowed a 3×3DDin size, pigmented, well circumscribed excavated lesion involving the centre of fovea with bared scleral vessels at the base and the retinal vessels undisturbed at the margin.Optic disc is normal with no evidence of pigment dispersion in the mid periphery.A systemic evaluation, including skull and chest x rays, complete blood count, erythrocyte sedimentation rate, antinuclear antibody,C-reactive protein, and VDRL test was noncontributory. Serological tests for cytomegalovirus, rubella, and herpes simplex were negative. A Toxoplasma IgG antibody test was negative.

Fig: 1



A) FUNDUS PICTURE OF RIGHT EYE SHOWING MACULAR COLOBOMA



B) FUNDUS PICTURE OF LEFT EYE SHOWING MACULAR COLOBOMA

OCT-helps to confirm the diagnosis of macular coloboma

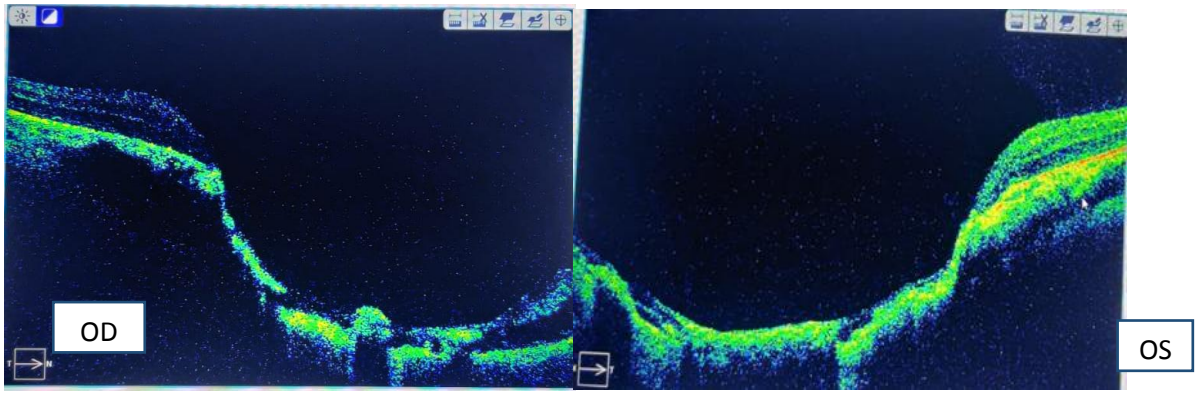


Fig2:- crater like depression in the macula with absence of RPE and choroid in the lesion

III. DISCUSSION

• Macular coloboma is a very rare birth defect with a prevalence of 0.5–0.7/10,000 live births. It is usually sporadic, although autosomal dominant or other inheritance patterns may be followed. It is thought to be caused by the failure of normal closure of the optic fissure between 5 and 7 weeks of development .Macular coloboma may result from intrauterine inflammation and can be associated with systemic developmental abnormalities. Mutation in BEST1 and RIMS1 have been identified.

• Mann classified macular coloboma into three types, namely pigmented macular coloboma, non-pigmented macular coloboma, and macular coloboma associated with abnormal vessels.

• Macular coloboma may be differentiated from, Best vitelliform macular dystrophy (BVMD) advanced cone-rod dystrophy (CORD),congenital toxoplasmosis macular scar,central areolar choroidal dystrophy .

• Macular coloboma, located at the posterior pole, can lead to retinal detachment leading to an increase risk of developing complete vision loss. Therefore, regular follow-up for patients with macular coloboma is indicated. During each follow up a dilated fundus examination should be done for the patient in order to elicit any breaks at the coloboma edges. Treatment of macular coloboma using laser photocoagulation is limited in its role in preserving the vision. patients with macular colobomas should be treated carefully for any refractive errors to maximize and optimize the patient's visual acuity.

IV. CONCLUSION

• Congenital coloboma is a rare ocular condition that leads to non-progressive decrease in visual acuity and if not followed up regularly may cause retinal detachment and vision loss. Optical coherence tomography is the method of choice in diagnosing macular coloboma while treatment should be done by laser photocoagulation if the coloboma is away from the macula with careful observation and follow-ups.

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