

CASE REPORT

OCULAR COLOBAMATA IN A FAMILY

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ABSTRACT: AIM: To identify the proportion of cases affected in a family with ocular colobamata. **MATERIAL:** Ocular colobomata present in a family **METHOD:** Visual acuity, Slit lamp examination, Ophthalmoscopy, Fundus Photography, B-Scan, Family members were examined. **RESULTS:** Family showed poor visual acuity, Iris and Choroidal Coloboma without optic disc involvement and normal corneal diameter. **CONCLUSION:** Ocular coloboma occurring in patients of my study is family and genetically determined.

KEYWORDS: Coloboma Iris, Coloboma Choroid.

INTRODUCTION: Coloboma is derived from the Greek "KOLOBOMA,"¹ meaning mutilated or curtailed. The malformation refers to a notch, gap, hole, or fissure in any of the ocular structures.^{2,3} coloboma encompasses any defect in the lids, iris, retina / uvea / choroid & / or optic nerve due to the faulty closure of optic fissure during 5th to 7th week of fetal life. The typical coloboma is caused by defective closure of fetal fissure.^{4,5} located in infero nasal quadrant & it may affect any part of globe traversed by fissure from iris to optic nerve. Complete evaluation of affected individual and their families is required in order to establish the etiology, prognosis, recurrence risk for each proband. Isolated colobomatus malformation in the absence of other systemic malformations, are most commonly inherited in an autosomal dominant manner, although autosomal recessive inheritance seems likely in some families.

We report herein families having ocular coloboma, without any systemic involvement.

CASE REPORT: A 50 year old female patient came to OPD of Princess Esra Hospital on November 2014 with complaint of Diminished vision in OD since 1 year. Her son aged 28 years came with the complaint of abnormally shaped pupils OU since childhood. Her daughter aged 31 years came with the complaint of diminished vision and abnormal shaped pupil in OS.

CASE 1:

Mother Visual acuity: OD - CF ½ mtr, OS - 6/18.

Slit Lamp Examination:

OU: Eye lids, conjunctiva, cornea - NAD

OD: Iris - notch in the infero nasal quadrant Pupil - key hole appearance, reacting to light.

Lens - IMSC Posterior segment - choroidal coloboma

OS: Iris - normal, pupil - R/R/R Lens - IMSC, Posterior segment – within normal Limits.

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Fig. 1: OD showing Ocular coloboma with IMSC, OS with in Normal Limit.

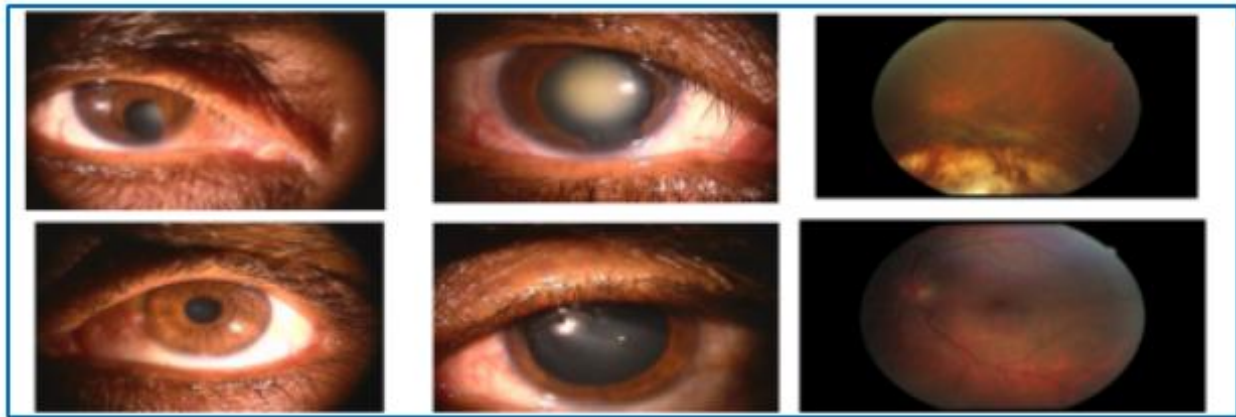


Fig. 1

CASE 2:

Son Visual acuity: OD - 6/6, OS - 6/6.

Slit Lamp Examination: OU eyelids, conjunctiva, cornea were normal.

OD showing in iris notch in the inferonasal quadrant. Small strand of mesodermal tissue bridging the notch, pupil showing key whole appearance reacting to light, lenticular coloboma with early cortical cataract, choroidal coloboma in the posterior segment.

OS showing in iris notch in the inferonasal quadrant, pupil showing key whole appearance reacting to light, lenticular coloboma with early cortical cataract, choroidal coloboma in the posterior segment.

Fig. 2: OU Ocular Coloboma with early cortical cataract.

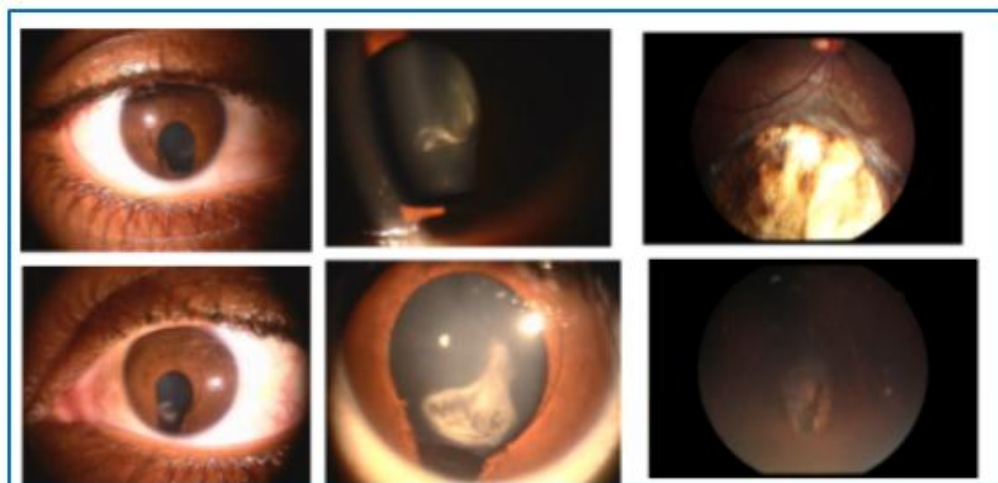


Fig. 2

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CASE 3:

Daughter Visual acuity: OD - 6/6, OS - 6/24 - BCVA - 6/18

Slit Lamp Examination:

OD: Eyelids, conjunctiva, cornea, Iris, Pupil, Lens -NAD, Posterior segment – WNL.

OS: Eyelids, conjunctiva, cornea NAD, Iris-notch in inferonasal quadrant, Pupil-key hole, Lens – cortical cataract. Posterior segment – choroidal coloboma.

Fig. 3: OS Ocular Coloboma.

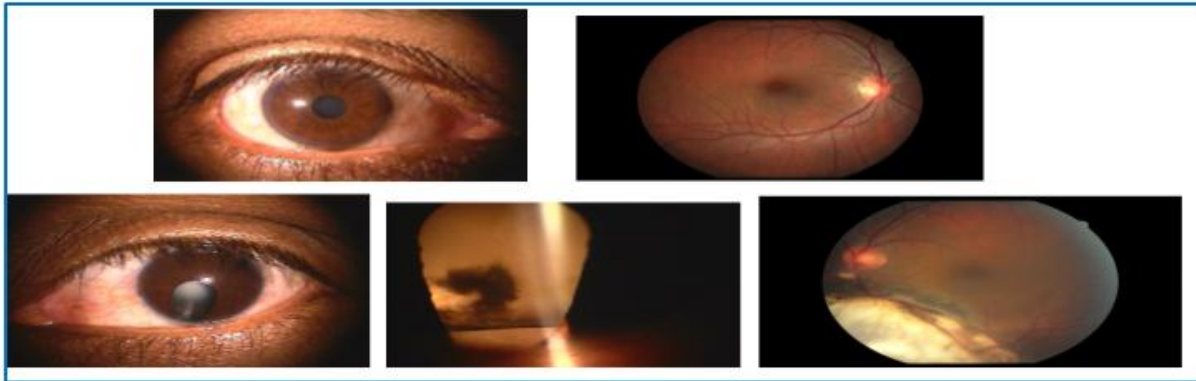


Fig. 3

TREATMENT: Case with severe lenticular opacities was treated with lens extraction and iol placement. Since the incidence of retinal detachment is high, so prophylactic laser photocoagulation is applied to all the three patients with Choridal Coloboma.

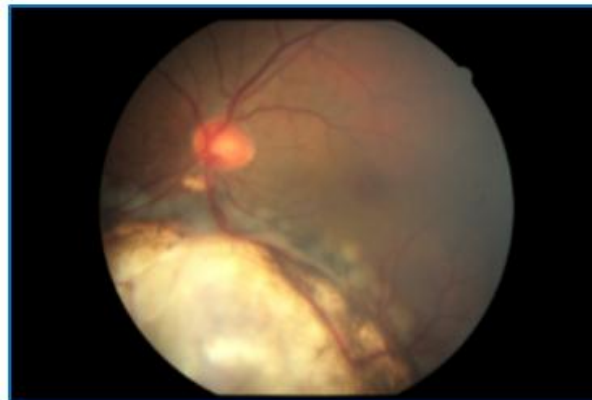


Fig. 4

DISCUSSION: Congenital ocular colobomata (From the Greek koloboma, meaning “mutilated” or “curtailed”) are caused by defects in embryogenesis. Typically, colobomata are clefts caused by absence of tissue in the inferonasal quadrant of the eye. The underlying aetiology of the phenotype is the failure of the ectodermal optic vesicle fissure to close. It occurs in 0.5 to 2.2

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cases per 10000 live Births. This leads to colobomata affecting one or more areas of the eye including the cornea, iris, ciliary body, lens, retina, choroid, and optic nerve. The typical, most frequently observed, ocular coloboma is seen in the inferonasal quadrant. A complete iris coloboma involves the pigment epithelium and stroma giving rise to the so-called "keyhole" pupil, which can be unilateral or bilateral. Although isolated iris coloboma is observed, it is often associated with colobomata in other parts of the eye. Chorioretinal coloboma, colobomata affecting the posterior segment of the eye can be unilateral or bilateral. If the fetal fissure fails to close posteriorly, then a coloboma affecting the retinal pigment epithelium (RPE), neurosensory retina, or choroid may occur. The defect is essentially a bare sclera with the overlying RPE, retina, or choroid missing. Typically occurring in the inferonasal quadrant. Usually, chorioretinal colobomata are asymptomatic despite significant upper visual field defects. It has been proposed that 23–42%.^{6,7} of cases can be complicated by retinal detachment and prevention is mainly by retinal lasers. Isolated colobomatus malformation in the absence of other systemic malformations, are most commonly inherited in an autosomal dominant manner, although autosomal recessive inheritance, x linked inheritance seems likely in some families.

Clinically most patients with Coloboma are asymptomatic. But complications such as retinal detachment and Cataract are associated with retinochoroidal Coloboma. Cataracts of multiple varieties are associated with coloboma including pigment deposits, sub capsular, cortical, anterior and posterior polar and total opacification. The treatment for lens Coloboma in severe cases is lens extraction with iol placement.^{8,4,9} and chorioretinal Coloboma is prophylactic laser photocoagulation to prevent retinal detachment.⁶

Our cases presented with decreased vision. On examination under slit lamp iris coloboma with lenticular opacities was seen. On fundus examination chorioretinal coloboma was seen. So, we have treated patient with severe lenticular opacities with lens extraction and iol placement. And patients with refractive error were treated with corrective glasses and cases having chorioretinal Coloboma were treated with prophylactic laser photo coagulation.

CONCLUSION: Ocular coloboma occurring in patients of my study is family & genetically determined which is rare.

There was wide variation in severity. In case presented to us mother is having uveal coloboma in right eye. Left eye was normal. Son was having both eyes uveal coloboma in both eyes. And daughter was having uveal coloboma in left eye.

Case having cataract in which visual function was sufficiently compromised was treated with cataract surgery, patient having retino choroidal colobomata was treated with prophylactic laser therapy along the edge of coloboma.

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