ORIGINAL RESEARCH PAPER

Ophthalomology

A CASE REPORT ON BOTH EYES IRIS COLOBOMA AND RETINOCHOROIDAL COLOBOMA INVOLVING DISC AND MACULA WITH EARLY CATARACT

KEY WORDS: iris, coloboma, cataract, visual acuity, macula, optic disc

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ABSTRACT

Purpose*:** A 22 year old male presented with diminution of vision in both eyes associated with ocassional pain in since childhood. On examination, visual acuity OD:6/60 and OS:Counting finger at 1.5m with both eyes no pinhole improvement. ***Materials And Methods: General physical examination revealed a moderately built and well nourished male, well oriented to time, place and person with no neurological deficits. Slit lamp examination of anterior segment of both eyes showed inferonasal coloboma of iris with pear shaped pupil;fundus examination revealed bilateral choroidal coloboma involving optic disc and macular lutea of the retina with early cataract. ***Result:*** Patient was managed conservatively with systemic painkillers and prophylactic laser delimitation of coloboma margins and Phacoemulsificaton with PCIOL implantation. ***Conclusion:*** This is a case of iris and retinochoroidal coloboma complicated with cataract managed with laser and cataract surgery to improve best corrected visual acuity.

INTRODUCTION

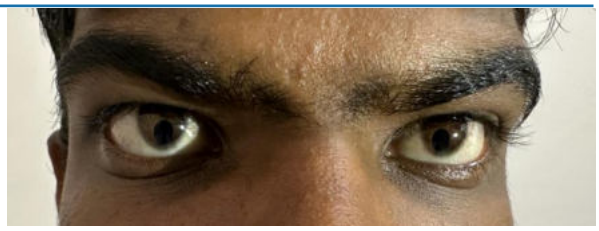
Coloboma is a Greek word that means mutilation.1 Ocular coloboma is a rare malformation which occurs as an isolated defect in healthy individuals or be part of a complex malformation syndrome of known or unknown etiology .[2,3,4]

Coloboma is generally classified as typical or atypical, depending on their location in the iris or fundus.Typical coloboma is the term used to describe the defects seen in the inferior/ infero -nasal part of the fundus that can be clearly attributed to defect in closure of embryonalfissure.5Similar defects seen elsewhere have been termed atypical coloboma.5Complete coloboma describes ocular defects of eyelids, cornea, lens, ciliary body , iris, zonules, choroid , retina and optic nerve.5

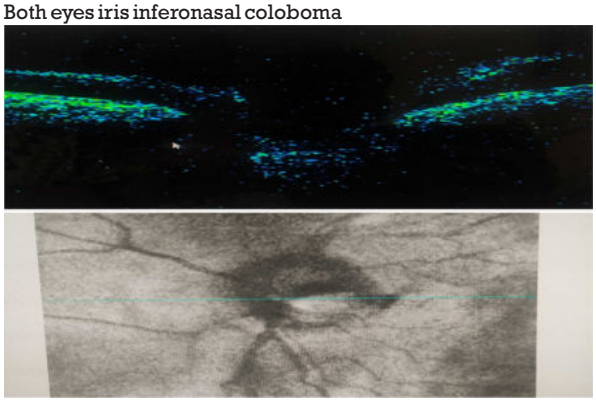
Sporadic coloboma is common, although several systemic abnormalities and syndromes have been described, for some of which genetic loci have been identified.5Coloboma is associated with several syndromes , CHARGE syndrome being the commonest. 5 Ocular colobomas are rare condition occurring in only 0.5 to 2.4 infants per 10.000 live births.6 The retinochoroidal colobomas present most frequently out of whole colobomas that accounting for 60-70% .6

MATERIAL AND METHODS

Descriptive single case report. A 22 year old male presented with diminution of vision in both eyes associated with occasional pain since childhood. On examination, visual acuity OD:6/60 and OS : Counting finger at 1.5m with both eyes no pinhole improvement. No history of trauma.No family history of similar complaints.General physical examination revealed a moderately built and well nourished male, well oriented to time, place and person with no neurological deficits. Slit lamp examination of anterior segment of both eyes showed inferonasal coloboma of iris with pear shaped pupil and early lens opacities indicating cataract formation. Fundus examination revealed bilateral retinochoroidal coloboma extending to the optic disc and macular lutea of the retina with early cataract Hypopigmented areas corresponding to the missing retinal layers and associated retinal thinning.No obvious retinal detachment was observed in both eyes.

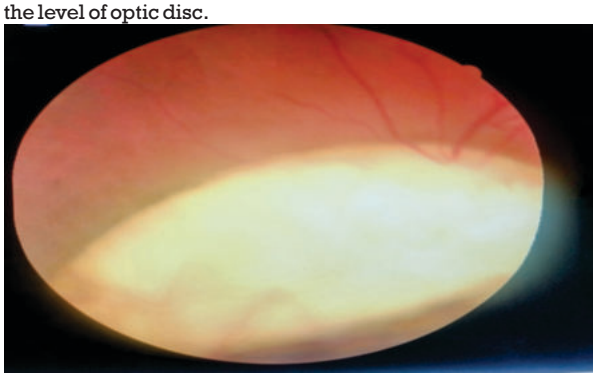


Both eyes iris inferonasal coloboma



Bilateral Inferonasal Coloboma Of Iris

OCT showing coloboma (disruption of outer retinal layers)at the level of optic disc.



Fundus image showing retinochoroidal coloboma Involving optic disc (not clear because of cataract)

RESULTS

Patient was managed conservatively with systemic painkillers and prophylactic laser delimitation of coloboma margins.Phacoemulsificaton with PCIOL implantation in left eye followed by right eye to manage cataract.Post-op visual acuity was OD:6/30 OS:6/36 and with pinhole OD:6/24 OS:6/30.

DISCUSSION

Chorioretinal colobomas are in many instances asymptomatic but often cause diminished visual acuity or distorted vision, especially when extending to the macula or optic disc.(7)The most common complications are retinal detachment and choroidal neovascularization.(7)Cataract surgery in these eyes can pose a challenge due to combination of microphthalmos and relatively hard lenses, resulting in increased risk of intra-operative complications.

CONCLUSION

This is a case of iris and retinochoroidal coloboma complicated with cataract managed with laser and cataract surgery to improve best corrected visual acuity. Coloboma is a complex disorder with a variable prognosis and requires regular examination to optimize visual acuity and to monitor for potential complications.

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