ORIGINAL RESEARCH PAPER
Volume - 12 | Issue - 08 | August - 2023 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

CLASSICAL FINDINGS OF CHORIORETINAL COLOBOMA - CASE SERIES

Ophthalmology

Dr. Ramya. B Final Year Postgraduate MS Ophthalmology, Tagore Medical College And Hospital , Chennai

Dr. Ravikumar. R Head Of Department, Professor, Department Of Ophthalmology, Tagore Medical College And Hospital , Chennai

Dr. Prasanthi Associate Professor

Dr. Premsai* Assistant Professor, Tagore Medical College And Hospital *Corresponding Author

ABSTRACT

Coloboma can develop as a result of abnormal fetal fissure closure in the fifth to sixth week of pregnancy. Coloboma is extremely uncommon, occurring in only 0.7 per 10,000 live births . We would like to present to you two such cases with varying age group .

KEYWORDS

Coloboma of iris, chorioretinal coloboma, fetal fissure abnormality, congenital

INTRODUCTION

Coloboma of the eye is a unique deformity that can develop as a single feature in a healthy individuals or as a component of malformations in a syndromes [2,3] .The uvea, lens, retina, Macula and optic nerve are all potential sites of ocular coloboma. There are three types of coloboma: typical, atypical and macular. The inferonasal position of a typical coloboma may be a result of the location of the optic fissure. Any location besides the inferonasal area may have an atypical coloboma. A pigmented abnormality in the macula characterizes the heterogeneous deformity known as macular coloboma[4].Coloboma can be associated with cataract, microphthalmos, anophthalmos, glaucoma and phthysis bulbi with visual impairment. Coloboma affects 0.5 to 2.4 newborns out of every 10,000 live births [4,16].A greater incidence ofcoloboma,60–70%, or 0.14% of the general population, was found in chorio-retinal colobomas. Parental consanguinity is connected with 28.8% of colobomas [13] .coloboma has variable prognosis which requires regular eye examination to help in better visual outcome and to prevent potential complication.

CASE 1

A 50 Female presented with complaint of defective vision in Right Eye for one year. No history of trauma or surgery to both eyes . Patient had no commodities. No history of any familial disorder. On Examination best corrected visual Acuity (BCVA) was Right Eye plano with 1\60 and LE with 6/24 respectively . On Anterior segment Examination RE pear shaped typical iris Coloboma present at 6 o clock position , pupil sluggishly reacting to light for both direct and indirect light reflex with Nuclear sclerosis grade I .Dilated fundoscopy relieved Normal size, shape,color of the disc, with healthy neuroretinal rim , cup disc ratio of 0.3:1 , vessels Normal in caliber Macula normal with chorioretinal Coloboma at 5 to 7 o clock position in inferior quadrant. Examination of left Eye revealed normal color and pattern iris , pupil sluggishly reaction to light both direct and indirect light reflex, lens - nuclear sclerosis of grade II, Fundus Examination revealed clear media with Normal size, shape,color of the disc, with healthy neuroretinal rim , cup disc ratio of 0.3:1 , vessels Normal in caliber Macula normal and FR +.

Diagnosis RE Typical complete Coloboma with iris and chorioretinal involvement with nuclear sclerosis of grade I, LE senile immature cataract with nuclear sclerosis of grade II

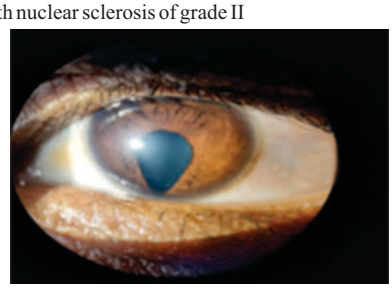

Image 1 Right Eye Iris Coloboma

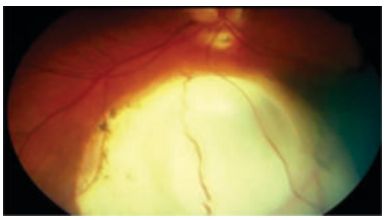

Image 2 Right Eye Chorioretinal Coloboma

CASE 2

A 5 year old boy came presented with complaint of decreased size of left Eye since birth and also complaints of diminished vision in both eye from birth .informant mother- reliable source .No history of preterm delivery , No history of mile stone delay, No history of trauma. No family history suggestive similar complaint in family members. On examination vision BCVA in RE 6/24, N6 and LE 6/60,N36 Respectively. RE Anterior segment iris Normal in color and pattern, pupil round 3mm reacting to both direct and indirect light reflex with clear len . Dilated fundoscopy Revealed a clear media with Normal size, shape, color of Disc with healthy neuro retinalrim , cup disc ratio of 0.3:1, vessels in Normal in caliber, inferior quadrant Choriorentinal Coloboma was present (sparing disc), Macula normal FR+

Left Eye anterior segment Examination Revealed microphthalmos with scleral thinning and microcornea with horizontal measurement 9 mm . Iris normal color and pattern , pupil 3mm round ,reacting to direct and indirect light reflex with clear lens and full and free Extraocular movements . Dilated fundus examination LE revealed chorioretinal Coloboma in inferior quadrant retina . Diagnosis Right Eye Chorioretinal Coloboma.Left Eye Choriorentinal Coloboma with microphthalmos. Patient was advised with low vision therapy.

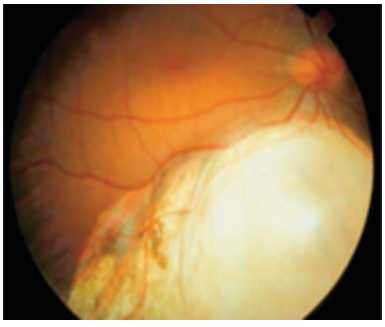

Image 3 Right Eye Chorioretinal Coloboma

DISCUSSION

Ocular Coloboma is a rare congenital malformation due to incomplete closure of embryonic fissure. Incidence of ocular Coloboma is 0.5 -2.2 cases per 10,000 live births [4].Incidence of Eyelid Coloboma is 0.2- 0.8

cases per 10,000 live births.[5]. In study by Kelly N.Nakamura et all prevalence of Ocular Coloboma is 1 in 2011 live birth with Anterior segment coloboma 36%, Posterior segment coloboma with 39% and 24% had both Anterior and posterior segment coloboma .coloboma can involve iris , zonules, ciliary body, choroid, Retina and optic nerve seperately or in combination with one and other. [6] ocular Coloboma can present in two extremes, it can cause nonfunctional eye by orbital cyst compression or it can have minimal involvement that hardly affect vision. Coloboma may present with defective vision gradual or sudden. Sudden onset vision loss is due to coloboma is associated with retinal detachment. Patient may leukocoria if it is large coloboma [7]. syndromes associated with Coloboma [7] are Isolated iris coloboma-Seckel syndrome 3 (14q24.3 (SCKL)), Meckel Gruber syndrome (17q22-q23 (MKS1)),Reiger's syndrome type 1 (4q25-q26 (PITX2))

Chorio-retinal coloboma±Iris coloboma±optic disc coloboma-Warburg micro syndrome 1 (2q21.3 (RAB3GAP)),Microphthalmia (14q24.3 (CHX10)),Congenital contractural arachnodactyly syndrome (Beal's syndrome) (5q23-q31(FBN2))

Isolated optic disc coloboma-Walker–Walburg syndrome (9q34.1 (POMT1)),Renal coloboma syndrome (10q24.3(PAX2))

Sporadic Coloboma: Coloboma can occur due environmental insult and due maternal causes such as eldery age group pregnancy, fetal alcohol syndrome .[8,9]

In Moseley M fields et all revealed Coloboma can lead to ambylopia in 1.6-3.5% and strabismus in 4 - 6% individuals [10,11]

Posterior segment coloboma are more vision threatening when it involves the optic disc than anterior segment coloboma . A study by Daufenbach et all described that Coloboma can lead to Retinal Detachment in 6% individuals.[12] Choroidal Coloboma can be associated with microphthalmos, high myopia, glaucoma , cataract and phthisis bulbi which can cause severe visual impairment . A study by O M Uhumwango revealed percentage of risk of retinal detachment in patient with prophylactic laser photocoagulation was lesser 2.9% than with untreated cases 24.1% [13] . Coloboma with retinal detachment has the worst prognosis and it's very much challenging for ophthalmologist to manage it due it's varying nature of size, location of retinal break when associated with other ocular anomalies such as microphthalmia, cataract, lens and iris coloboma. Hotta k et al tried to seal the Coloboma with cyanoacrylate glue but results was not satisfactory [14]. scleral blucking was tried during earlier days but prognosis was poor in cases with retinal detachment and Coloboma [15, 16]. Nowadays surgeons prefer to do pars plana vitrectomy with or without vitreous substitute.

CONCLUSION

Coloboma in early age group are unaware of their visual problem in most of circumstances so they present at later age with advanced disease where it would have left to ambylopia, Retinal Detachment with or without macular involvement. Earlier rehabilitation of young individuals may help in reduction of Ambylopia and strabismus with help of recent advances such us low vision aids, patching of eyes. Posterior segment coloboma is complex to deal with and visual outcomes are guarded even with this recent advances.

Conflict Of Interest : Authors declare no conflict of interest
Source Of Funding : Nil

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