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Case Series

Case Series of IRIS and Choroidal Coloboma with Other Ocular and Systemic Associations

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Abstract:

Objective: To explore the various ocular and systemic associations in patients with iris and choroidal coloboma for better understanding of clinical implications and management strategies.

Methods: This is a prospective study done over 1 year in patients with coloboma. Detailed history followed by complete ophthalmic examination including anterior and posterior segment, a comprehensive review of medical records and evaluation was performed to identify associated anomalies.

Results: A total of 8 patients with coloboma were included in this case series with a median age of 26 years. Out of 8 patients, 2 were males and 6 were females. 75% of cases presented with bilateral coloboma while the rest 25% of cases exhibited unilateral involvement. 88% of patients had visual acuity from <6/60 to PL+. Various ocular associations identified in these patients include microphthalmia in 38%, cataracts including congenital and posterior subcapsular cataracts in 50% cases, nystagmus in 25% and posterior staphyloma in 38% cases. Only 1 patient had systemic association such as charge syndrome.

Conclusion: Iris and choroidal coloboma are rare congenital ocular anomalies that arise due to incomplete closure of the optic fissure during embryonic development. This case series reveals a diverse spectrum of ocular and systemic associations in patients with iris and choroidal coloboma. Early detection and intervention for associated conditions can significantly impact the long-term visual and overall outcomes of the affected individuals.

Keywords: Coloboma, Ocular and Systemic Associations, Congenital Cause of Blindness.

Introduction

Coloboma are rare congenital anomalies resulting from defective closure of the optic fissure during embryonic development. Closure of the embryonic fissure occurs during the second month of embryonic development. Embryonic fissure is usually located in the inferonasal quadrant, and it is the most common site for coloboma. Most colobomas result from the failed closure of the embryonic or choroidal fissure during the 5th to 7th week of fetal life. [1,2] It can be unilateral or bilateral and may be sporadic or inherited and present with systemic associations in some cases. [3] Colobomas of the iris or ciliary body result from failures of complete anterior closure, while colobomas of the choroid, retina or optic nerve result from failures of posterior closure. [4] Colobomas can be isolated or may be associated with other ocular disorders such as microphthalmia [5] cataract, nystagmus or systemic associations like CHARGE syndrome. [6,7] Retinal detachment and cataract are the most common complications associated with retinochoroidal coloboma. Coloboma of the posterior pole is associated with an increased risk for retinal detachment with occurs in 23-42% of patients. [8]

Aim & Objectives

To explore the various ocular and systemic associations in patients with iris and choroidal coloboma for better understanding of clinical implications and management strategies.

Material and Methods

Institutional ethical committee approval was obtained for this study. This is a prospective hospital based observational study done over a period of one year. Patients with holes or defects in the ocular structures were identified and examined for the presence of iris and choroidal coloboma.

Demographic data regarding age and sex were collected for each patient with coloboma. Detailed history regarding previous medical history and family history of coloboma were noted. Complete ophthalmic examination is done starting with visual acuity, anterior segment examination with slit lamp to determine the location of coloboma and other ocular abnormalities were noted. Posterior segment evaluation was done to look for the presence of choroidal coloboma. A comprehensive review of medical records of the patients with coloboma was performed to identify systemic associations such as charge syndrome etc.

Here we describe two cases of coloboma from our study emphasizing visual acuity, ocular features, and systemic associations.

Case 1

A 7-year-old female child presented to outpatient department with visual impairment in both eyes. In the right eye, her best corrected visual acuity was measured at 6/60, and in the left eye, there was no perception of light. Notably, the pupil in the right eye briskly reacted to light, while in the left eye, pupil was not reaction to light. A slit lamp examination of the right eye revealed a keyhole defect in the inferonasal part of the iris, indicative of coloboma, and a dilated fundus examination confirmed the presence of choroidal coloboma. In the left eye, the slit lamp examination showed a similar keyhole defect in the inferonasal part of the iris, suggesting coloboma, along with the additional finding of a congenital cataract. B-scan ultrasonography of left eye demonstrated the presence of posterior staphyloma. This was associated with nystagmus in both eyes. Notably, no other systemic associations were identified.

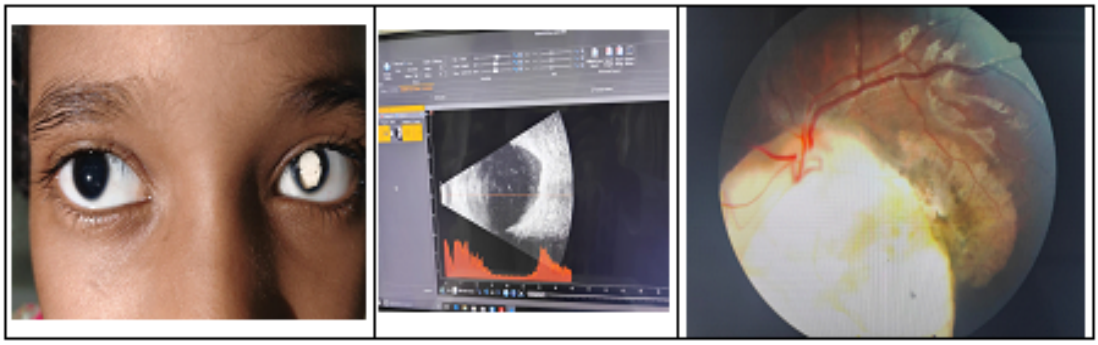

Figure 1.

Case 2

A 22-year-old female presented to outpatient department with defective vision in both eyes. Upon examination, her best corrected visual acuity was 6/36 in right eye and counting fingers at two meters in the left eye. Pupils were briskly reacting to light without any relative afferent pupillary defect. A detailed slit lamp examination showed a defect in the inferior part of the iris, strongly indicative of coloboma in both eyes. Posterior segment examination revealed the presence of choroidal coloboma in both eyes. Notably, the patient's past medical history revealed that she had hearing defects since childhood. Furthermore, she had been diagnosed as having coronary atherosclerotic heart disease with sub aortic peri membranous ventricular septal defect (VSD). At the age of 10 years, she had undergone surgical intervention for the closure of the ventricular septal defect. Polydactyly was also noted upon further examination.

Figure 2.

Results

The results offer a comprehensive analysis of the prevalence and characteristics of the condition. In our study, a total of eight patients with coloboma were analyzed. This detailed examination yields crucial insights into the demographic, clinical findings and ocular and systemic features associated with coloboma, aiding in better understanding of the disease.

Age Wise Distribution

The demographic profile in our study reveals that there were two patients in the age group of 10-20 years, 21-30 years, and 41-50 years. Only one patient each was in the age group 0-10 years and 31-40 years as shown in table. The median age group was found to be 26 years among the eight patients analyzed.

Table 1

Age	Number of Patients
0-10	1
11-20	2
21-30	2
31-40	1
41-50	2
Age Wise Distribution	
Gender	**Frequency**	**Percentage**
Male	2	25
Female	6	75
Gender		
Laterality	**Frequency**	**Percentage**
Unilateral	1	12
Bilateral	7	88
Laterality of Coloboma		

Most of the patients i.e., seven (88%) out of eight patients had ocular associations other than coloboma. Only one out of eight patients had systemic associations such as CHARGE syndrome. The various ocular features other than coloboma in our study include microphthalmia in 38%, various cataracts including congenital cataract and posterior subcapsular cataracts in 50%, nystagmus in 25% cases, posterior staphyloma in 38% and strabismus in 50% of cases as shown in table.

Table 2: Ocular Associations of Coloboma

Association	Percentage
Microphthalmia	38
Cataract	50
Nystagmus	25
Posterior staphyloma	38
Strabismus	50

Particularly notable is the high incidence of severe visual impairment, with 88% of cases having best corrected visual acuity of less than 6/60. Two (25%) patients had involvement of anterior segment only while the remaining six (75%) patients had involvement of both anterior and posterior segment respectively.

Discussion

This case series yields crucial insights into the demographic, clinical, ocular, and various systemic features associated with coloboma. During our study period, we encountered only eight patients who were clinically diagnosed with coloboma. A population-based study done at Mayo clinic reported the diagnosis of coloboma in 33 children during 40-year study period. [2] Clarke [9,10] and his colleagues (1954) found 12 cases of typical colobomata in a series of 500 congenital ocular abnormalities. Ocular coloboma due to failed closure of embryonic fissure occurs in 0.5-2.2 cases per 10000 live births. [3] The reason for low prevalence in our study mainly could be due to short duration.

The study's demographic profile reveals a median age of 26 years among the eight patients analyzed. This median age suggests that coloboma significantly impacts individuals in their prime years, underlining the importance of early diagnosis and management. A cross-sectional hospital-based study done by Anthony Vipin Das et al also reported that the most common age group at presentation was individuals in the second decade of life, comprising 2198 patients (23%). [11]

The distribution of genders, characterized by a ratio of 25% males to 75% females, highlights a significant imbalance in the occurrence of coloboma within the studied population. In the Bell [12] (1932) study, no significance sex incidence was observed, as evidenced by an equal distribution of cases with 52 males, 52 females, and 7 cases of unrecorded sex out of 111 cases.

The predominance of bilateral cases (88%) in this study is a crucial finding. This high rate of bilateralism highlights the necessity for comprehensive ocular assessments in coloboma patients, as the condition often affects both eyes, potentially leading to symmetrical visual impairment. This exceeds the documented rates of bilateral involvement in coloboma patients in Hungary, [13] which is reported to be 47.5%. The elevated occurrence of bilaterality in our study could be attributed to instances where unilateral cases go unnoticed by the patients, possibly because the other eye compensates with better vision.

Out of the total, two patients (25%) exclusively displayed involvement in the anterior segment, whereas the remaining six patients (75%) exhibited engagement in both the anterior and posterior segments, respectively. In the study conducted by Bermejo et al., 55 cases of coloboma were reported, with 47% specifically affecting the iris, 40% presenting as chorioretinal coloboma, and the remaining 13% categorized as unspecified. [14]

The study's focus on ocular and systemic associations provides valuable insights into the broader impact of coloboma. Ocular associations were present in 87% of cases, with conditions such as microphthalmia, cataract, nystagmus, posterior staphyloma, and strabismus being reported. Gregory-Evans et al. and Chang et al. have authored outstanding articles that offer a thorough compilation of reported associations between coloboma and systemic syndromes, along with the identified gene loci or chromosome aberrations.

In this study, microphthalmia was present in 38% of cases of coloboma. Colobomatous microphthalmos is the condition wherein failure in the closure of the embryonic cleft is associated with undue smallness of the eye. [10] In cases of unilateral coloboma, the other eye may be microphthalmic and deformed. [12] The study also sheds light on the rare occurrence of systemic associations in coloboma, evidenced by the single case of charge syndrome identified.

Conclusion

The analysis of coloboma cases provides critical insights into the demographic, clinical, and ocular characteristics of the condition. The findings emphasize the importance of comprehensive ocular examinations, recognition of associated conditions, and the need for targeted interventions to manage visual impairment effectively. The study's implications reach beyond clinical practice, opening avenues for further research into the etiology and management of coloboma.

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