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A Comprehensive Clinical Description of Patients with Retino-Choroidal Coloboma, Presenting to a Tertiary Care Hospital in Eastern Uttar Pradesh

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Abstract

Introduction and Objectives: Typical Retinochoroidal [RC] coloboma is caused by a defective closure of the embryonic Fissure and can have varying presentations, differing from person-to-person and eye-to-eye. This study aims to identify the demographic and clinical profile of patients with RC coloboma presenting to a tertiary care teaching hospital in rural Eastern Uttar Pradesh.

Methodology: A retrospective cross sectional study, conducted between 26th April 2022 and 3rd May 2023, wherein the data was collected from the electronic medical record of registered patients presenting to the Vitreo-retinal OPD of Regional Institute of Ophthalmology, Sitapur.

Results: A total of 509 patients were included in the study. The mean age of presentation was 24.73 years, with almost equal. Out of the total patients, 256 patients (50.29%) had bilateral RC coloboma, while 253 patients (49.70%) had unilateral. The mean vision was 0.85 and 0.98 log MAR in right and left eye respectively. Both, the disc and macula, were involved in 65.61% patients, only macula was involved in 1.76% patients and only the optic disc was involved in 2.36% patients. In 154 patients (30.25%), both the optic disc and macula were spared. Other pathologies accompanying RC Coloboma were retinal detachment (16.90%), bilateral cataract (2.75%), unilateral cataract (17.09%), phthisical globe (2.94%), nystagmus (1.77%), anophthalmos (3.14%) etc.

Conclusion: Retinochoroidal coloboma has a wide spectrum of presentation. The visual compromise can be due to the coloboma itself or due to its complications. A better understanding of the presentation would help in a superior management with promising results.

Keywords: Retinochoroidal Coloboma; Coloboma; Iris Coloboma; RC Coloboma

Abbreviations

RC: Retino-Choroidal; VR: Vitreo-Retinal; OPD: Outpatient Department; BCVA: Best Corrected Visual Acuity.

Introduction

Coloboma is derived from a Greek term "koloboma" that stands for a defect or a mutilation [1]. A Typical Retinochoroidal

[RC] coloboma is a rare eye malformation caused by defective closure of the embryonic Fissure [2]. The occurrence of coloboma can be either be sporadic, hereditary or may be associated with chromosomal abnormalities. When caused by chromosomal abnormalities, ocular colobomata can be associated with systemic abnormalities and syndromes, for some of which genetic loci have been identified [3]. The eyes with ocular colobomata can have varying presentations, and may frequently be associated with various ocular anomalies, like cataract, microphthalmia, anophthamia et cetera [4,5]. The fundal coloboma can present with diminution of vision, depending upon degree of involvement of optic disc or macular involvement, or upon complications viz-a-viz rhegmatogenous retinal detachment (RRD) [6,7]. This study aims to identify the demographic and clinical profile of patients with RC coloboma presenting to a tertiary care teaching hospital in Eastern Uttar Pradesh.

Materials and Methods

The current study is a retrospective analysis of electronic medical records of all patients with RC coloboma presenting to the Vitreo-retinal outpatient department of Regional Institute of Ophthalmology, Sitapur, India from 26th April 2022 to 3rd May 2023. Data regarding the patients' age, gender, best corrected visual acuity, presenting complaints, anterior and posterior segment details and the advised treatment were taken. Patients whose vision cannot be assessed were excluded from the study.

Results

Out of 1, 60,924 patients presenting to Regional institute of Ophthalmology, Sitapur, 509 patients (0.32%) were found to have either unilateral or bilateral RC coloboma. The average age of presentation was 24.73 years (Range =5-86 years), consisting of 52.82% males and 47.15% females. Figure 1 shows the age-wise gender distribution of RC coloboma patients.

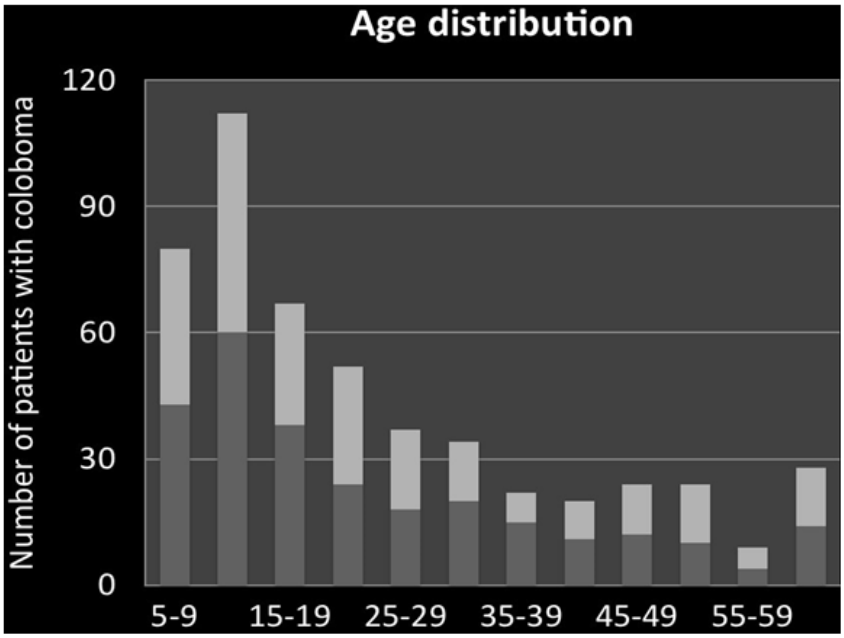

Figure 1: Showing the gender-wise distribution of patients with RC coloboma presenting at different age groups- males (dark) and females (light).

Bilateral RC colobomata were present in 256 patients (50.29%) and unilateral colobomata were present in 253 cases (49.70%). Figure 2 shows the ratio between bilateral and unilateral cases presenting with RC coloboma.

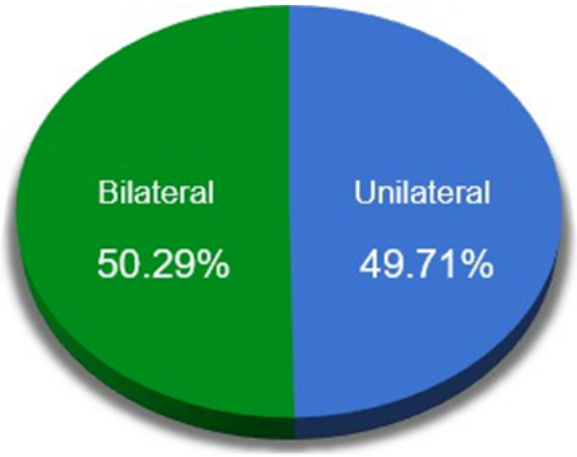

Figure 2: Showing the ratio between bilateral and unilateral cases presenting with RC coloboma.- bilateral (green), unilateral (blue).

BCVA (log MAR) ranged from 0.00 to no perception of light, with 85.07 % eyes having visual acuity of equal to or worse than 0.0 and better than light perception in right eye and 81.14 % eyes having visual acuity of equal to or worse than 1.0 and better than light perception in left eye. The mean vision in right eye was 0.85 log MAR, while that in left eye was 0.98 log MAR. Both, the optic disc and macula, were involved in 65.61% patients. Only macula was involved in 9 patients (1.76%) and only the optic disc was involved in 12 patients (2.36%). In 154 patients (30.25%), both the optic disc & macula were spared. Figure 3 shows RC Coloboma shows sparring both disc and macula (left) and RC Coloboma involving the optic disc (right) and Figure 4 shows the proportions of different fundus Findings in patients with RC coloboma.

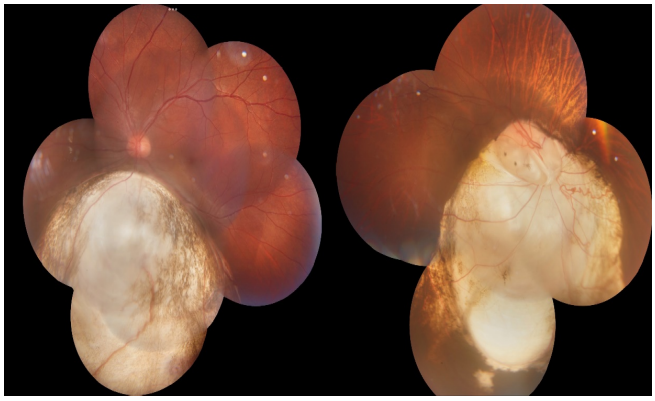

Figure 3: Showing RC Coloboma shows sparring both disc and macula (left) and RC Coloboma involving the optic disc only (right).

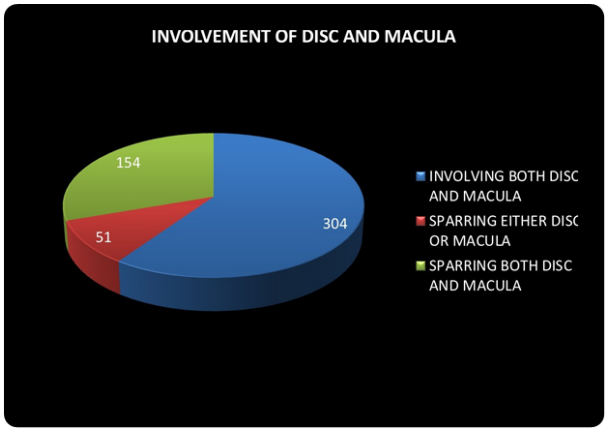

Figure 4: Showing proportion of various RC coloboma patients involving both disc and macula (blue), sparring either disc or macula (red) and sparring both disc and macula (green).

Associated ocular anomalies were present in about one-third eyes (32.61%), with cataract being the most common, presenting in a total of 87 eyes (17.09%) unilaterally and in 14 patients bilaterally (2.75%). Other accompanying ocular anomalies observed were retinal detachment (16.90%), prephthisical globe (1.96%), phthisical globe (2.94%), nystagmus (1.77%), anophthalmos (3.14%) and posterior staphyloma (2.94%). Figure 5 shows the proportion of various ocular anomalies associated with RC Coloboma.

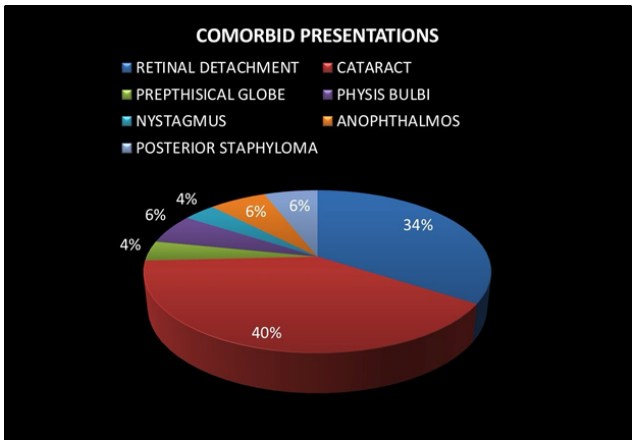

Figure 5: Showing the proportion of various ocular anomalies associated with RC Coloboma.

Discussion

Ocular coloboma is a rare congenital malformation that can present along a wide spectrum of anomalies- ranging from iris coloboma to clinical anophthalmos [2,7]. The present

study was done with the aim of defining a comprehensive clinical analysis of patients with RC coloboma presenting to a tertiary care hospital in Eastern Uttar Pradesh. For the study, a retrospective analysis of electronic medical record system was done, in which, out of all the outpatient department, RC coloboma patients presenting to VR OPD of Regional Institute of Ophthalmology, Sitapur, were assessed.

The prevalence of RC coloboma patients noted in our study was 509 out of the 1, 60,924 patients, making the prevalence to be 0.32%. A study by George A, et al. [8] says that the prevalence of RC coloboma varies anywhere between 0.5 to 2.6 per 10,000 births. Studies by Stoll C, et al. [7,9] in 1992 and 1997 show the prevalence of RC colobomata to be 0.7 and 1.4 respectively in 10,000 eyes. Another study by Bermejo E, et al. [10] in 1998 says that the prevalence of RC coloboma in newborns was found to be 1.96 in 1, 00,000 eyes. In 2021, Kohli G, et al. [11] claimed in their study that the prevalence of colobomata in eyes undergoing cataract surgery was found to be 0.085 %. A study by Nakamura KM, et al. [12] done in Minnesota 2021 says that the live birth prevalence of RC colobomata was surprisingly high, that is, 1 in 2077 population, in pediatric population (<19 years of age).

Out of all the patients diagnosed with RC coloboma, most male: female ratio was almost equal, with males being 52.82% while females being 47.15%. RC coloboma has no direct gender predilection. In 2011, Nakamura KM, et al. [12] also show in their study that out of 33 patients of diagnosed ocular coloboma, 17 (52%) were males and 16 (48%) were females. However, studies by Pagon RA in 1981 and Lingam G, et al. [2,3] in 2021 show that colobomata associated with syndromes may have some gender predilection, for example, only females are affected in Aicardi syndrome (X linked dominant). Similarly, in colobomata associated with Goltz's facial dermal hypoplasia (X linked dominant), most patients affected are females. A similar variation was also noted in the study by Gopal L, et al. [4] in 1996, which noted 25 (62.5%) males and 15 (37.5%) females, out of 40 patients under study.

The mean age presentation in our study was found to be 24.73 years (Range =5-86 years). Gopal L, et al. [4] shared nearly similar findings in their study in 1996, wherein they noted the age at presentation to be 6 to 60 years (average, 21.58 years). Apart from some screening studies of new-borns by Stoll C, et al. 1997 and Bermejo E, et al. in 1998, or study on pediatric population by Nakamura KM, et al. in 2011 (median age of presentation=3.9 months), a big number of typical RC coloboma patients may go undiagnosed or may not even present to the hospital [7,10,12]. The study by Barnard S, et al. [13] also claims under-ascertainment of posterior segment coloboma unaffected vision in some cases. The biggest reason cited for this by a study by Olsen TW, et al. [5] was the diminution of vision based on the degree of fundal involvement. Best corrected visual acuity, noted using log MAR scale, ranged from 0.00 to no perception of light, with 85.07 % eyes having visual acuity of equal to or worse than 0.0 and better than light perception in right eye and 81.14 % eyes having visual acuity of equal to or worse than 1.0 and better than light perception in left eye. The mean vision in right eye was 0.85 log MAR, while that in left eye was 0.98 log MAR. In the study by Nakamura KM, et al. [12] done in 2011, BCVA was less than 20/60 in 9 of the 28 patients (32%).

The most important etiology of poor visual acuity is the degree of fundal involvement. In our study, we found that both, the optic disc and macula, were involved in 65.61% patients. Only macula was involved in 9 patients (1.76%) and only the optic disc was involved in 12 patients (2.36%). In 154 patients (30.25%), both the optic disc & macula were spared. A study done by Gopal L, et al. [4] on the status of optic disc in fundal colobomata categorized these eyes in 6 categories, based on their optic disc status-

- Normal disc outside fundus coloboma (27.8%).
- Disc outside the fundus coloboma and abnormal (10.4%).
- Disc outside the fundus coloboma and independently colobomatous (8.9%).
- Disc within the fundus coloboma and normal (5.0%).
- Disc within the fundus coloboma and colobomatous (44.3%).
- Disc shape not identified but blood vessels seen emanating from the superior border of the large fundus coloboma (2.9%).

Thus, Gopal L, et al. [4] claims a presence of a linear relationship between poor vision and degree of fundal involvement in RC colobomata. In 2003, Berk AT, et al. [14] also found similar results in their study - optic disc and choroid were involved in 18 eyes, an isolated disc coloboma was present in 5 eyes while a normal optic disc was present in 4 out of total 30 eyes included in the study. Nakamura KM, et al. [12] also showed in their study that out of 13 patients of diagnosed RC coloboma, 4 patients had only macula involved, 8 patients had only optic disc involved and 1 patient had both disc and macula involved. However, according to Gopal L, et al. [4] in 1996, degree of fundal involvement is not the only cause of diminution of vision in RC coloboma patients. Other factors like amblyopia, retinal detachment, cataract, etc, might be few of the other reasons.

The present study shows equal results as far as the laterality is concerned- with Bilateral RC colobomata were present in 256 patients (50.29%) and unilateral colobomata were present in 253 cases (49.70%). The study by Lingam G, et al. [3] states that the involvement can be unilateral in 33-47.5% of cases and bilateral in remaining patients. Similarly studies

by Vogt G, et al. 2005 and Barnard S, et al. in 2012 found unilaterality in 50% and 40% cases respectively [13,15]. On the contrary, the study by Nakamura KM, et al. [12] states that the prevalence of unilateral RC coloboma noted was 67%. This was hypothesized to be due to reporting or referral bias, thus favoring bilaterality.

RC colobomata are known to be associated with variable degrees of other ocular abnormalities. The current study found out the prevalence of retinal detachment (16.90%), prephthisical globe (1.96%), phthisical globe (2.94%), nystagmus (1.77%), anophthalmos (3.14%) and posterior staphyloma (2.94%) in eyes of RC coloboma patients. In their study in 2003, Berk AT, et al. [14] found that among 9 patients, 14 eyes were microphthalmic and 9 eyes of 6 patients had micro cornea. Also, retinal detachment was present in 1 patient. Similarly, the study by Bermejo E, et al. [10] suggested that anophthalmia/microphthalmia + coloboma was the frequent combination of eye defects and was found in 19 cases. The study by Gopal L, et al. [4] discussed in detail about the various associated ocular anomalies coexisting with RC coloboma. According to their study, the overall incidence of retinal detachment was 42.8%. Microphthalmos was also significantly prevalent, seen more often in the more severe anomalies. They also found prevalence of nystagmus in patients primarily having disc involvement and bilaterally poor vision. Phthisis bulbi was found in 1 eye and complicated cataract in 3 eyes. In 2014, Uhumwangho OM, et al. [16] also found significant phenotypic variations among 198 patients in their study, with the prevalence of retinal detachment being 17.6%, micro cornea being 45.1%, nystagmus being 41.5%, strabismus being 21.2%, and microphthalmos being 19.1%.

Conclusion

The variability of various phenotypic variations and different aspects of clinical profile parameters of eyes with RC coloboma is quite diverse. It's a rare congenital abnormality with almost equal gender predilection, unless syndromic. The mean age of presentation is 24.73 years with the mean visual acuity being 0.85 log MAR and 0.98 log MAR for right and left eyes respectively. Degree of fundal involvement was the biggest factor cited for the diminution of vision, other factors being amblyopia, retinal detachment or cataract. Both unilateral and bilateral cases shared equal shares of the cohort. RC colobomata are also associated with variable degrees of other ocular abnormalities, viz-a-viz, retinal detachment, prephthisical globe, pthisical globe, nystagmus, anophthalmos and posterior staphyloma. Hence, a better understanding of other clinical associations in patients with RC coloboma makes us look for them more diligently which favours further prognosis.

Conflict of Interest

There are no financial interests or conflicts of interest that exist with this study.

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