ORIGINAL RESEARCH PAPER

Ophthalomology

CLINICAL PROFILE OF RETINOCHOROIDAL COLOBOMA IN PEDIATRIC PATIENTS AND ITS ASSOCIATED COMPLICATIONS IN A TERTIARY EYE CARE HOSPITAL IN INDIA

KEY WORDS:
Retinochoroidal coloboma, paediatric, retinal detachment, cataract

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ABSTRACT

Background– Retinochoroidal coloboma (RCC) is an underdiagnosed condition in pediatric patients, which is subsequently associated with various ocular and systemic co-morbidities. Lack of awareness and delayed treatment may exacerbate the condition. **Objective –** to study the clinical profile of retinochoroidal coloboma in fifty pediatric patients (<20yrs) and its associated complications in a tertiary hospital in India **Materials and Methods–** in an institution based retrospective cross-sectional study (approved by the institutional ethics committee), fifty pediatric patients aged <20years diagnosed with retinochoroidal coloboma (RCC) were evaluated for their demographic and clinical profile. **Results–** out of the 79 eyes examined, around 50% eyes had grade II or grade III RCC. The most common complications associated were cataract (60%), microphthalmos (26%) and rhegmatogenous retinal detachment (26%). Systemic abnormalities were detected in 6% patients. **Conclusion–** early diagnosis of RCC is essential to preserve visual acuity in pediatric patients and prevent ocular complications.

INTRODUCTION:

Retinochoroidal coloboma (RCC) is characterized by the congenital absence of part of the retinal pigment epithelium (RPE) and choroid, caused by the defective closure of the embryonic fissure, which normally develops gradually between the sixth and seventh weeks of foetal life. They account for 60-70% of the total incidence of colobomas and reported to be 0.14% of the general population.[1] RCC can be sporadic or genetically transmitted as an autosomal recessive, autosomal dominant or X-linked trait. Uhumwangho and Jalali[1] reported parental consanguinity in 28.8% of the 198 patients in their series. RCC may also be associated with other ocular pathologies such as cataract, microphthalmia, nystagmus and anophthalmia.[1]

The severity of **visual disability** is highly variable and is dependent on many factors; including the size of coloboma, extent of macular or optic nerve involvement, and associated anomalies of the globe such as microphthalmos, microcornea, retrobulbar cysts and nystagmus. These clinical findings coexist nearly in 90% of eyes.[2] Vincent et al.[3] reported that the visual acuity is generally poor and 87% of eyes with colobomas had a visual acuity worse than 20/200. Ocular involvement can range from small colobomas with isolated chorioretinal involvement, to large colobomas affecting the iris, choroid, retina and optic nerve.[4] Increased coloboma excavation is linked with higher incidence of retinal breaks and rhegmatogenous retinal detachment (RRD).[5,6,7]

RCCs present as a prominent de-pigmented white zone within the fundus which is usually located in the inferonasal quadrant, that is the latest part of closure of the embryonic fissure.

But they have a significantly heterogenous clinical appearance according to extent of involvement and affected part of different anatomical areas such as macula and optic nerve.

Uhumwangho and Jalali[1] reported 7 types of colobomas according to **Ida Mann classification**:
Type 1—coloboma extending above the anatomic disc,
Type 2—coloboma extending up to superior border of disc,
Type 3—coloboma extending below the lower border of disc,
Type 4—coloboma involving the disc only,
Type 5—coloboma present below the disc with normal retina above and below the coloboma,
Type 6—pigmentation present in the periphery,
Type 7—coloboma involving only the periphery
It is reported that the both retinal and choroidal vessels patterns are directly affected by the size and area of the coloboma.[8]

Retinal detachment is the most common **complication** of RCC and the rate of rhegmatogenous retinal detachment (RRD) is significantly higher than that in the general population. RRD related to RCC have been reported in 2% in the general population and up to 8% in children under age of 10yrs.[9] According to optical coherence tomography (OCT) analysis, RRD in eyes with RCC have been found to be most common secondary to breaks in the abnormally thin inner retina; ICM and communication between both the sub-ICM space and subretinal space.[1,3,10,11]

According to Rishi et al.[12] report in eyes with optic disc involvement, fluid may enter subretinal space through the colobomatous defect in the optic nerve tissue. Other associated ocular anomalies include microphthalmos, microcornea, amblyopia, strabismus and cataract.[13]

METHODS AND METHODOLOGY:

I. Study Design:
Institution based retrospective case control study

II. Place Of Study:
Paediatric patients aged less than 20 years of age attending the outpatient department of ophthalmology in a tertiary eye care centre in India.

III. Period Of Study: January 2021 – January 2022 (One year)

IV. Study Population:
A sample size of 50 patients, who attended the out-patient department within the study period, and were diagnosed with retinochoroidal coloboma (RCC) for the first time and hadn't been treated elsewhere prior to current visit.

V. Parameters For Assessment:
Thorough ocular examination, which included best corrected visual acuity after adequate cycloplegia, anterior segment evaluation with slit lamp bio-microscopy and posterior segment evaluation using indirect ophthalmoscopy and USG/OCT as and when required.

Systemic evaluation included evaluation of milestones, CVS, CNS, genitourinary and skeletal systems and appropriate paediatric workup to rule out congenital anomalies.

VI. Approval From Institutional Ethics Committee obtained for the present study.

RESULTS:

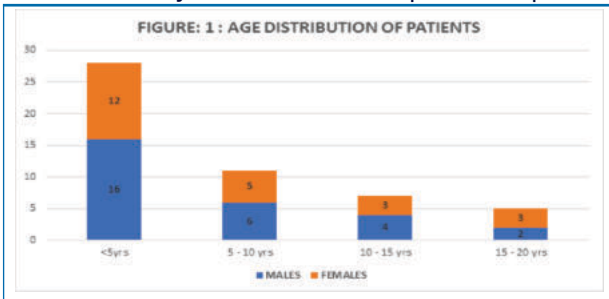

Figure: 1: Demographic Distribution Of Patients

Out of the 50 patients evaluated, 28 patients (56%) were male. Most of the patients were below 5yrs of age (28 patients; 56%); 10 patients (20%) were in the age group 5 – 10 years, 7 patients (14%) in the age group 10 – 15 years and 5 patients (10%) in the age group 15 – 20 years.

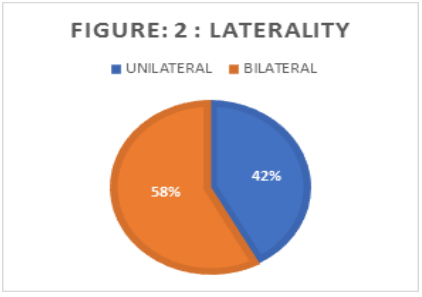

Figure: 2: Laterality Of Presentation

Out of the 50 patients, 29 patients (58%) had bilateral retinochoroidal coloboma, as compared to 21 patients (42%) with unilateral disease.

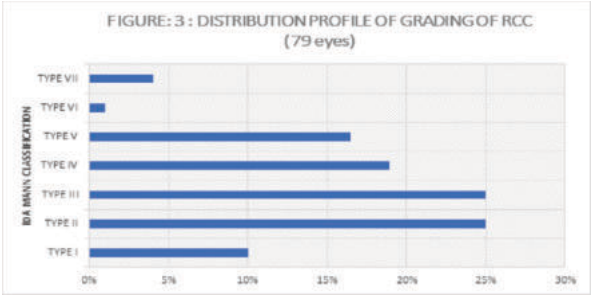

Figure: 3: Distribution Of Rcc Grading (according To Ida – Mann Classification)

Out of the 79 eyes examined (in 50 patients), the highest proportion of RCC was of Grade II (20 eyes, 25%) and Grade III (20 eyes, 25%). 15 eyes (19%) were of Grade IV, while 13 eyes (16%) were of Grade V. The least common was Grade VI, with only 1 patient (1%) of the presentation.

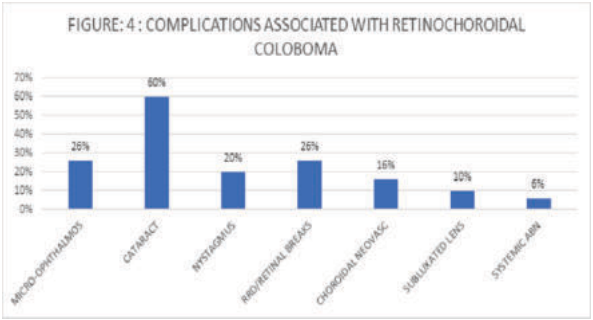

Figure: 4: Clinical Profile Of Complications Associated With Rcc

Ocular and systemic complications were more common with Ida Mann Grade I and II ($p<0.001$). The most common ocular complication was cataract (47 eyes; 60%), followed by microphthalmos and RRD with or without retinal breaks (20 eyes each; 26%). 16 eyes had nystagmus (20%), 13 eyes (16%) had choroidal neovascularization and 8 eyes (10%) had subluxated lens. Only 4 eyes / 2 patients (6%) had associated systemic anomalies.

DISCUSSION:

Retinochoroidal coloboma (RCC) encompasses a group of similar findings of congenital tissue defects, due to maldevelopment and incomplete closure of the embryonic fissure. It appears clinically as a prominent whitish or yellow-white 'excavated' zone, usually in the inferonasal quadrant of the ocular fundus. The lesion may have rudimentary retinal tissue, with a few ectatic blood vessels over the sclera. The neurosensory retina continues as an intercalary membrane (ICM) in the area of the coloboma.

Uhumwangho OM et al[1] (2014) studied 335 eyes of 198 pediatric patients with chorioretinal colobomas, wherein bilaterality was observed in around 69.7% of patients and 53.5% patients were male. Parental consanguinity was documented in 28.8%. The mean age of presentation was 7.8 ± 4.75 years. 46.9% eyes were graded as type II Ida Mann coloboma. 87.2% eyes were associated with ocular anomalies, with iris coloboma being the most common in 238 (71%) eyes, microcornea (45.1%), nystagmus (41.5%), strabismus (21.2%), and microphthalmos (19.1%).

Daufenbach et al[4] (1998) studied eighty-six eyes in pediatric patients and estimated a prevalence of around 8.1% in the general population, as opposed to prior estimates of 23% to 43%. Ocular involvement was variable and the combined prevalence of retinal or choroidal detachment was 10.4% of patients and 8.1% of affected eyes. 38% of patients were diagnosed with associated systemic abnormalities.

The **present study** concluded with 56% male patients, with 58% associated with bilaterality. Both type II and type III Ida Mann colobomas were of highest prevalence, about 25% each. 56% of the patients were <5years of age, as opposed to previous study results of mean age of 7-8yrs.[1] Ocular complications were more common in types I and II colobomas, with cataract being the most common ocular complication (60%). Systemic associations were present in 6%.

Nakamura KM et al[6] described ocular coloboma in thirty-three pediatric patients, mean age being 3.9 months of age, with 67% unilaterality. 58% patients had associated ocular disorders, including amblyopia (33%) and strabismus (30%). 67% had associated systemic abnormalities, with abnormal development in 36% and CHARGE syndrome in 12%.[6,7]

The most important complication of chorioretinal colobomas is retinal detachment with a prevalence ranging from 2.4% to 47.5%.[7] Incidence of retinal detachment and retinal breaks are higher in these patients due to the thin and undifferentiated intercalary membrane (ICM) overlying the retinal defect. Hussain et al., however, reported that retinal breaks occurred outside the coloboma in 5 of 15 eyes with colobomatous retinal detachments.[14] These authors hypothesized that there may be an abnormal vitreoretinal interface not limited to the area of the coloboma and its margin in colobomatous eyes. **The current study found the incidence of RRD to be around 26%.** Colobomatous retinal detachments should be repaired surgically despite a guarded visual prognosis, particularly in children with bilateral chorioretinal colobomas, given the risk of future vision loss in the contralateral eye. Some authors have even suggested prophylactic laser in all patients with chorioretinal colobomas, given the high risk of retinal detachment in this population.[4,14]

An association has also been demonstrated between chorioretinal colobomas and choroidal neovascularization (CNV) occurrence at the temporal margin of the coloboma through defects in the Bruch membrane.[15] **The current study**

found a prevalence of CNVM in 16% of the patients. CNV in colobomatous eyes can spontaneously resolve or lead to progressive worsening of visual acuity. When associated with worsening vision, CNV can be treated with anti–vascular endothelial growth factor (anti-VEGF), focal laser photocoagulation, or photodynamic therapy.[14,15]

Chorioretinal colobomas can occur in isolation or can be associated with systemic syndromes, such as CHARGE (coloboma, heart abnormalities, anal atresia, renal abnormalities, genitourinary abnormalities, eye abnormalities) syndrome, Goldenhar syndrome, Rubinstein-Taybi syndrome, trisomy 18, 4p syndrome, basal cell nevus syndrome, Aicardi syndrome, congenital rubella, Walker-Warburg syndrome, and Joubert syndrome.[4,16] The current study evaluated the incidence of systemic abnormalities at around 6%. Patients with bilateral chorioretinal coloboma or unilateral chorioretinal coloboma plus one other systemic abnormality should be referred for genetic and chromosomal testing for systemic disorders.

CONCLUSION:

Ocular coloboma may be inherited in a multitude of ways, and show overt symptoms initially, which may progress insidiously, to result in debilitating vision loss in later stages of life. Pediatric cases are underdiagnosed and undertreated, which may cause irreversible loss of vision. The present study concluded the maximum incidence in <5years age group, bilateral being the more common presentation. Types II and III were the most common presentations, with most cases being associated with cataract, microphthalmos and RRD. Prophylactic laser and timely surgical intervention for RRD is necessary for vision preservation. 6% of the patients were found with systemic associations. Hence, screening for systemic abnormalities is essential for bilateral retinochoroidal colobomas or in patients with unilateral retinochoridal coloboma, with one additional systemic symptom.

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