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# Bilateral choroidal colobomas: A rare case report and literature review

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## Abstract

Ocular colobomas are congenital gaps in the ocular anatomy, namely choroidal colobomas. It happens when the embryonic fissure fails to fuse, leaving a chronic defect. Measuring just 0.14% of eye patients, choroidal colobomas might be inherited or spontaneous, making genetic identification difficult. Although colobomas typically cause no symptoms and are discovered by accident, they can sometimes cause vision loss. Numerous abnormalities are linked to colobomas, and their consequences—such as retinal detachment—call for specialized treatments such as preventive laser therapy. We report the case of an eighteen-year-old female patient with bilateral choroidal coloboma, presenting with iris coloboma and impaired vision. This study emphasizes how crucial it is to identify colobomas as soon as possible for the best possible care.

**Keywords:** Coloboma, Fetal Fissure, Retinal Detachment, Microcornea, Prophylactic Laser Retinopexy

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## Introduction

The word coloboma comes from the Greek, meaning curtailed or mutilated. In ophthalmologic usage the word refers to any "notch, gap, hole, or fissure in any of the ocular structures whether congenital or acquired.[1]
An unusual ocular anomaly, orbital colobomas arise from a disruption in the normal merging of the fetal fissure within the optic cup. The lower part of a number of anatomical systems, such as the choroid, retina, ciliary body, lens, iris, optic nerve, and retinal pigment epithelium, are typically affected by these defects.[2] A choroidal coloboma is identified by the congenital absence of a portion of the retinal pigment epithelium and choroid.[3] Clinically, it is identified as a noticeable white area in the fundus, typically situated inferiorly and slightly toward the nasal side.[4]

Several studies have been conducted to evaluate its frequency. The prevalence rates of colobomas vary, according to population-based research; they range from 3.7 per 100,000 based on a national registry in Hungary to 8 per 100,000 in Scotland.[5]

Colobomas can be classified as either typical or atypical based on the affected structure. Colobomas that involve the inferior or inferonasal part of the fundus are considered typical, whereas those occurring in other areas of the eye are termed atypical. Atypical colobomas, unlike typical ones, are not associated with chorioretinal or optic disc coloboma, as they do not indicate deficiencies in embryonic fissure closure.[1]

## Case report

An 18-year-old female who presented with at ophthalmology OPD with chief complaint of blurring of distant vision for 5 years which has been increasing for 4-5 months. She had used specs since the 4 years of age. There was no history of itching, pain, and redness. She had no systemic abnormalities. No similar defect in siblings.

On examination the visual acuity of both eyes had choroidal colobomas. On ocular examination, iris coloboma and cortical cataract were seen in right eye. Ocular mobility was present bilaterally in all directions. On ophthalmoscopy cup disk ratio was 0.6:1 on both eyes but no glaucomatous changes were present. On non-contact tonometry (NCT), intraocular pressure was 17 mmHg in both eyes, Table 1.

On fundoscopy with dilation of the pupil, choroidal coloboma type 3 was seen in the right eye, and choroidal coloboma type 5 and type 7 in the left eye, Figure 1.

On the Humphrey field analyser, the Visual field index (VFI) on the right eye was 50% with a single upper scotoma visual field defect, and on the left eye, VFI was 88% with a single upper peripheral visual field defect, Figure 2.

The final diagnosis was right eye iris coloboma with choroidal coloboma type 3 with micro-cornea with cortical cataract with myopia and left eye choroidal coloboma type 5 and 7 with myopic astigmatism.

**Table 1. Visual acuity of both eyes in 18-y lady with bilateral choroidal coloboma**

| Eye | Unaided | With pin hole | With power glass |
|---|---|---|---|
| RE (OD) | 6/36 | 6/24 | 6/24<br>(-3.75 DC x 180°) |
| LE (OS) | 6/36 | 6/6 | 6/6<br>(-1.00 DSPH/ -1.00 DC x 180°) |

**Figure 1. Fundus photos of eyes showing, (a) Right eye choroidal coloboma type 3, (b) Left eye choroidal colobomal type 5 and type 7**

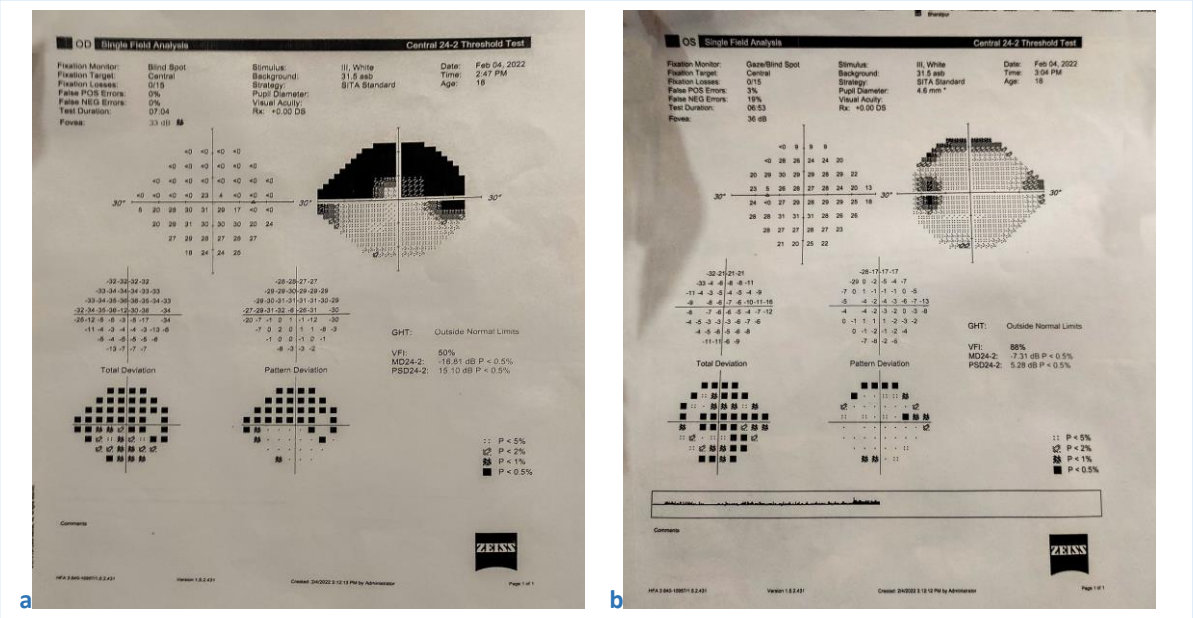


**Figure 2. Humphrey field analyzer (HFA), visual filed index (VFI), (a) Right eye VFI was 50% with a single upper scotoma visual field defect, (b) Left eye VFI was 88% with a single upper peripheral visual field defect**

## Discussion

This case is of an 18-year-old female presented with isolated bilateral choroidal coloboma progressive blurring of vision.

Ocular coloboma is a rare condition. Choroidal coloboma is an infrequent finding occurring in 0.14% as reported in a large series of eye patients.[4] This rare ocular malformation occurs because of a defect in the fusion of the fetal fissure of the optic cup during the fifth week of development.[2]

Colobomas are unilateral (33–47.5% of cases) or bilateral (symmetric or asymmetric). According to the study, 36% of patients had single anterior segment involvement (iris and ciliary body), 39% had isolated posterior segment involvement (chorioretinal and optic nerve), and 24% of cases had involvement of both.[6] Here in the present case, the patient had both segment involvement in the right eye and isolated posterior segment involvement in the left eye.

Choroidal coloboma has been classified into 7-types according to Ida Mann Classification.[3]

- Type 1: coloboma extending above the anatomic disc
- Type 2: coloboma extending up to the 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

In the present case, on fundoscopy examination with dilation of the pupil, choroidal coloboma type 3 was seen in the right eye and choroidal coloboma type 5 and type 7 were seen in the left eye.

Colobomas are either sporadic, inherited (known or unknown gene deficiencies), and related to chromosomal abnormalities.[6]

Identifying the genetic basis of colobomas has proven challenging. Syndromic colobomas, as seen in conditions like CHARGE syndrome, Treacher-Collins syndrome, and Patau syndrome, often have discernible genetic defects. However, in instances of non-syndromic colobomas, over 70% do not exhibit genetic defects.[5] The sporadic appearance of colobomas may be attributed to environmental factors leading to intrauterine insult. Risk factors for coloboma include Vitamin A deficiency, maternal diabetes, hypothyroidism, and medication like thalidomide, carbamazepine, hydantoin, alcohol, etc. during pregnancy.[6] No such environmental risk factors were observed in the present case.

Choroidal colobomas frequently manifest with diverse symptoms. In many instances, parents or pediatricians identify the condition due to noticeable anomalies, such as a small eyeball, pronounced nystagmus, strabismus, or a distinct iris coloboma. Prolonged impaired vision can stem from uncorrected refractive errors, amblyopia, or involvement of the fovea. In this particular case, the patient exhibited an iris coloboma in the right eye and reported a heightened blurring of vision over the last 4-5 months, having used corrective glasses since the age of 4 years. Additional symptoms may involve sudden vision deterioration, often attributed to conditions like retinal detachment (RD) or the development of a subfoveal choroidal neovascular membrane. Asymptomatic cases exist, especially with small colobomata not affecting the disc or fovea unless complicated by RD. A child's yellow/white pupillary reflex (leukocoria) or complicated cataract can alert parents.[6] This patient also has a cortical cataract in the right eye for which no intervention was required. Cataract surgery has greater complications in coloboma.[6]

Choroidal coloboma is frequently presented together with other anomalies like iris coloboma, microphthalmos, microcornea, cataract, lens coloboma, and optic pit. This patient also had microcornea. Other associated birth defects and complications include heart failure, hearing impairment, and intellectual disability.[2]

Retinal detachment (RD) occurs more frequently in posterior segment colobomas due to retinal breaks.[5] Approximately 2.4% to 47.5% of eyes affected by choroidal coloboma may experience the development of retinal detachment.[6] Breaks in Bruch's membrane at the margin of the colobomas frequently result in choroidal neovascularization (CNV).[7]

The treatment strategy for retinal detachment is individualized, with retinopexy being the primary intervention.[5] Advances in vitreo-retinal surgical techniques have driven the evolution of its treatment. Initially, scleral buckling was commonly used but has largely been replaced by the more prevalent vitrectomy approach.[6]

Prophylaxis laser therapy of the entire coloboma margin may be done to prevent RD.[6] Without prophylactic laser treatment, there is a 10.6 times higher risk of rhegmatogenous retinal detachment (RRD).[5] Since our patient had no retinal detachment and so no intervention was required but she was advised for prophylactic laser coagulation therapy and referred to Bharatpur eye hospital, Chitwan.

Colobomas are a rare eye problem that might not show clear signs at first but can later cause serious issues leading to sudden vision loss.

Recognizing it early is crucial to avoid complications. The challenge is that people with colobomas often don't have symptoms, so they might not go for checkups until after they've already lost vision due to problems like retinal detachment. Regular eye checkups can help find potential cases, but there aren't specific programs to screen for retinal detachment yet. So, it's important to increase awareness and create screening plans for that. Prophylactic laser retinopexy can lower the chances of retinal detachment.

## Conclusion

This case report of an 18-year lady who has been using specs since the age of 4 years, had no other symptoms, and presented with progressive blurred vision of four months. She was finally diagnosed with a rare condition of bilateral choroidal colobomas.

Because of higher risks of retinal detachment, she was advised for prophylactic laser coagulation therapy and referred to Bharatpur eye hospital, Chitwan. Coloboma can be asymptomatic. Awareness programs and regular eye checkups for timely diagnosis and treatment should be considered to prevent serious complication and vision loss.

## Author contribution

Concept, design- PD, HR; Literature review: All; Draft manuscript- All; Final manuscript and accountability of the work- All.

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## Conflict of interest

None

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## Supplementary material

The data and supplementary material that support the findings of this study are available from the corresponding author upon reasonable request.

## Consent

All the appropriate patient consent has been taken in verbal and written the form. The patient gave consent for her eye check-up images and other clinical information to be reported in the journal. The patient was assured that due efforts would be taken to conceal the identity, but anonymity cannot be guaranteed.

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