Scientific Research Publishing

Open Journal of Ophthalmology, 2025, 15(1), 1-6
https://www.scirp.org/journal/ojoph
ISSN Online: 2165-7416
ISSN Print: 2165-7408

Ocular and Palpebral Colobomas: About Three Cases at Iota

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How to cite this paper: Sangaré, R.D., Guirou, N., Ba, K., Konipo, A., Sissoko, M., Sidibé, F.T. and Touré, A.I. (2025) Ocular and Palpebral Colobomas: About Three Cases at Iota. *Open Journal of Ophthalmology*, **15**, 1-6.
https://doi.org/10.4236/ojoph.2025.151001

Received: July 15, 2024
Accepted: January 20, 2025
Published: January 23, 2025

Abstract

Coloboma is due to a defect in the closure of the colobomic cleft generally occurring between the 5th and 7th week of embryonic life. Coloboma can affect the adnexa and/or the eyeball. We report the iconography of two cases of ocular colobomas and one case of eyelids coloboma at the IOTA CHU. **Case 1:** This was a 32-year-old patient who consulted for decreased visual acuity in the left eye. The clinical examination found visual acuity of 10/10 in the right eye and hand movement in the left eye. Biomicroscopic examination revealed an iris and chorioretinal coloboma encompassing the papilla. **Case 2:** This was a 3-year-old child. Who consulted for eyelid malformation since birth. On ophthalmological examination, there was a bilateral coloboma of the inner half of the upper eyelid, associated with an underlying symblepharon and left eye corneal dystrophy. Our course of action consisted of a release of the symblepharon followed by a blepharoplasty which allowed a good anatomical and functional restitution. **Case 3:** This was a 15-year-old patient who consulted for bilateral visual acuity loss measured by counting fingers at 1m in the right eye and counting fingers at 3m in the left eye. The biomicroscopic examination found a bilateral papillary coloboma associated with a bilateral progressive cataract, an old detachment of the left retina. The etiological assessment was unremarkable. **Conclusion:** Coloboma is a congenital condition that can affect all parts of the eye. We reported three cases of colobomas, one palpebral, one iris and chorioretinal, and finally, one papillary associated with other ocular pathologies.

Keywords

Coloboma, Congenital, Unilateral, Bilateral

1. Introduction

Coloboma is a rare condition. Van Duyse defined it in these terms: the term

congenital coloboma applies to malformations of certain parts of the eye. Their configuration and location give a characteristic appearance to these anomalies. These are slits (iris), lacunae (chorio-retina), the normal tissue being absent, aplastic or replaced by connective tissue; they are changes in shape (optic nerve) relating, for all these anomalies, to a pathological process during intrauterine life. These malformations are usually located in the meridian corresponding to the level of the fetal slit, either directly below, or in a slightly more internal meridian (typical colobomas). They can be located in any other meridian [1].

It is due to a defect in the closure of the colobomic cleft generally occurring between the 5th and 7th week of embryonic life [2]. The prevalence of coloboma is 0.5 to 0.7 per 10,000 births [2].

The occurrence of coloboma can be sporadic, hereditary, caused by known or unknown genetic defects or associated with chromosomal abnormalities. Ocular coloboma is more often associated with systemic abnormalities when they are caused by chromosomal abnormalities [3].

Colobomas may be present in one or both eyes and, depending on their size and location, can affect a person's vision. Colobomas affecting the iris, which results in a "keyhole" appearance of the pupil, generally do not lead to vision loss. Colobomas involving the retina results in vision loss in specific parts of the visual field. Large retinal colobomas or those affecting the optic nerve can cause low vision, which means vision loss that cannot be completely corrected with glasses or contact lenses [4].

Forms affecting the posterior segment have a poor prognosis [5].

Cases of coloboma have been reported in the literature. The most frequent colobomas are chorioretinal [6].

We report cases of iris and chorioretinal colobomas and a bilateral eyelid coloboma.

2. Case Presentation

CASE 1: This was a 32-year-old patient, with no particular medical, surgical or family history, who consulted for decreased visual acuity in the left eye. The clinical examination found visual acuity without optical correction rated at 10/10 in the right eye and saw hand movements in the left eye. The biomicroscopic examination of the right eye was normal. Examination of the left anterior segment found a "keyhole" shaped iris with the most rounded part being superior. At the level of the posterior segment, a chorioretinal coloboma was noted exposing the sclera occupying 2/3 of the retina including the papilla (**Figure 1** & **Figure 2**).

CASE 2: This was a 3-year-old child who consulted for bilateral eyelid malformation since birth. On ophthalmological examination, there was a bilateral coloboma of the inner half of the upper eyelid, associated with an underlying symblepharon and corneal opacification in the left eye (**Figure 3**). We performed a release of the symblepharon (by excision of the eyelid that covered the upper part of the cornea by approximately 5 millimeters (mm). Followed by blepharoplasty

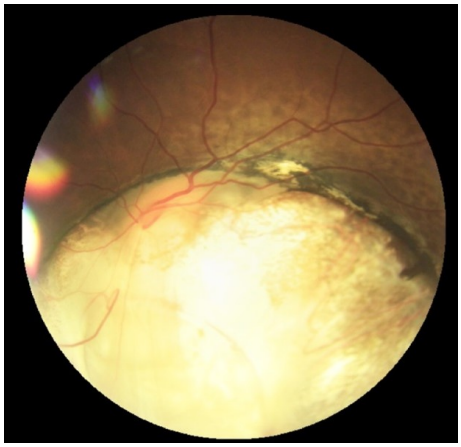

Figure 1. Chorioretinal coloboma encompassing the optic disc.

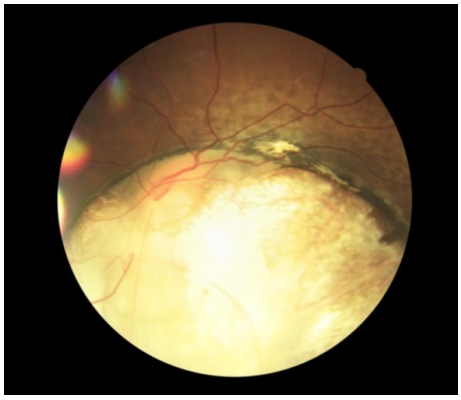

Figure 2. Chorioretinal coloboma encompassing the optic disc.

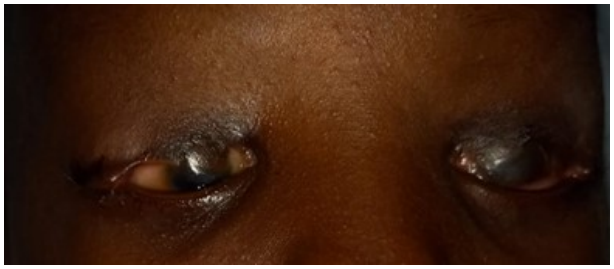

Figure 3. Bilateral palpebral coloboma with symblepharon.

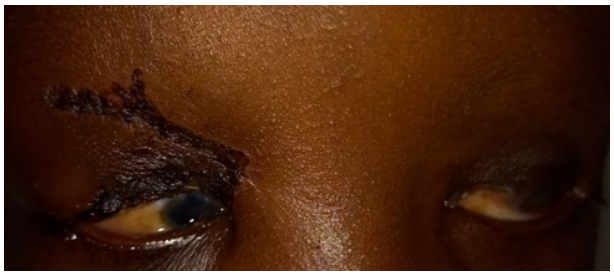

Figure. 4. Symblepharon release with graft on the coloboma of the right eye.

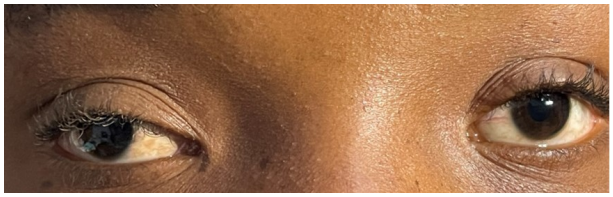

Figure 5. Microphthalmia and OD exotropia.

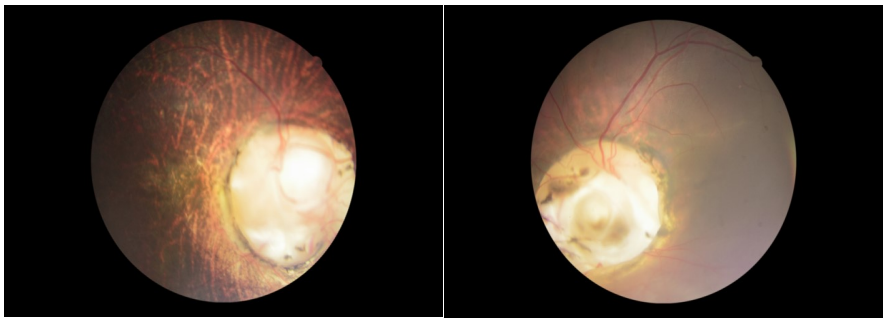

Figure 6. Bilateral papillary coloboma.

(a graft was taken from the skin of the upper eyelid and then sutured to the colobomatous part). This suture was done with 6/0 Vicryl. The postoperative course was simple. The blepharoplasty allowed good anatomical and functional restitution (**Figure 4**).

CASE 3: This was a 15-year-old patient with no particular medical, surgical or family history who consulted for bilateral visual acuity loss for several years. Visual acuity was measured at 1/20 in both eyes. On external examination, microphthalmia and exotropia were noted in the right eye (**Figure 5**). On biomicroscopy, there was an evolving cataract in both eyes. On examination of the posterior segment, a bilateral papillary coloboma was found (**Figure 6**) and a retinal detachment in the left eye. The biological assessment aimed at etiology, looking for Toxoplasmosis, Rubella and fetal-maternal syphilis, were negative. Optical coherence tomography (OCT) was inconclusive because visual acuity was too low. The patient is being treated for low vision.

3. Discussion

The occurrence of coloboma can be sporadic, hereditary, caused by known or unknown genetic defects or associated with chromosomal abnormalities. Coloboma can affect different structures of the eyeball and therefore we can find clefts that are located in the iris, the choroid, the retina, the lens, the eyelid or even the optic nerve [4]. In our series of cases no etiological research was done, due to lack of financial means.

At the level of the iris when the damage is complete, the missing surface is of variable importance. It results in a more or less extensive iris defect in the lower nasal with an appearance of pupillary deformation classically called a "keyhole" [5]. This is the case in our first observation.

In chorioretinal involvement, the coloboma is in fact initially a retinal coloboma and it is the absence of retinal development that results in the absence of development of the choroid. It appears on examination of the fundus in the form of a yellowish white territory, of variable size in the inferonasal territory which can go from the periphery to the papilla [7]. The iris coloboma can be isolated or associated with severe ocular anomalies such as microphthalmia [7]. In our third case, the coloboma is associated with microphthalmia.

Coloboma can be unilateral, as is the case in our first observation. Diallo S *et al.* at the IOTA University Hospital in 2018 found a bilateral case of chorioretinal coloboma. In our second and third observations, the coloboma is bilateral.

Eyelid colobomas are surgically treated, as they risk causing corneal ulcers [1]. Our second observation benefited from blepharoplasty with release of the symblepharon. There were no postoperative complications. The eye was anatomically and functionally restored.

Congenital colobomas are due to a failure of the fetal fissure to close. The diagnosis is made by MRI. Due to lack of financial means, our patients could not have an MRI. No genetic analysis was done in our observations. Because these examinations are expensive in our context. No other cases of coloboma have been diagnosed in the respective families of our patients.

4. Conclusions

Coloboma is a genetic condition. It can be associated with pathologies that can jeopardize the patient's visual prognosis.

We reported two cases of ocular colobomas and one case of palpebral coloboma. The first case was a unilateral iris and chorioretinal coloboma.

The second case was a bilateral palpebral coloboma.

The last case is a bilateral papillary coloboma associated with a bilateral progressive cataract and an old detachment of the left retina.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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