Case report of a patient with bilateral optic nerve coloboma

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Abstract
Coloboma is a congenital defect that can affect the anterior and posterior segment of one or both eyes and may cause visual impairment or can be associated with the rare but serious CHARGE (Coloboma of the eye, Heart defects, Atresia of the Nasal choanae, Retardation of growth and/or development, Genitourinary malformation Ear abnormalities and/or deafness) syndrome.

We present the case of a 10-year-old girl from a rural eye center in Kenya, who reported to the hospital for the first time with a smaller left eye and poor vision since birth. Her uncorrected visual acuity (UCVA) was 20/250 in the right eye (RE) and 20/400 in the left eye (LE). Examination revealed 15-degree esotropia in the LE, anisometropia, and bilateral coloboma. Systemic examination was normal. Best-corrected visual acuity (BCVA) was 20/60 in the RE with a refraction of +5.75/-0.50 × 112° and 20/150 in the LE with +4.50/-1.25 × 120°. Imaging with a Zeiss Clarus 700 fundus camera confirmed a larger chorioretinal coloboma in the LE, with no signs of retinal detachment. Genetic testing was not available. The patient was provided with spectacles and advised to have serial fundus photographs taken every six months at our center.

This case highlights the need to ensure the early diagnosis of coloboma and to assess for potentially life-threatening associations, such as CHARGE syndrome.

Key words: Optic nerve, coloboma

Background

Coloboma is a rare congenital ocular defect resulting from incomplete closure of the embryonic fissure, leading to the absence of specific eye tissues. It can affect both the anterior segment (eyelids, iris, lens, ciliary body, and zonules) and the posterior segment (choroid, retina, and optic nerve). In posterior colobomas involving the optic disc or choroid and retina (1), visual outcomes are influenced by the presence of foveal anomalies or retinal detachment, underscoring the need for regular fundoscopic examinations and serial imaging. Notably, there is no surgical intervention available for optic disc coloboma (1).

Bilateral optic disc coloboma is associated with poorer visual outcomes compared to unilateral cases. It is essential to differentiate optic disc coloboma from similar conditions, such as optic disc pit and morning glory syndrome. A thorough systemic evaluation is recommended to rule out CHARGE syndrome, a serious congenital disorder with an incidence of approximately 1 in 10,000 live births (2). The syndrome includes coloboma, heart defects, atresia choanae, growth retardation, genital abnormalities, and ear abnormalities.

This report presents the case of a 10-year-old girl from a rural eye centre in Kenya who exhibited significant ocular abnormalities. By detailing her clinical presentation and examination findings, we aim to underscore the importance of early diagnosis and appropriate management of coloboma and its associated conditions.

Case Presentation

A 10-year-old girl from rural Kenya presented at the Tenwek Mission Hospital for the first time with history of a smaller left eye and poor vision since birth. She did not have a family history of congenital ocular conditions or any other congenital anomalies. Both of her parents were healthy, and

they did not report any known genetic disorders affecting members of their families. The patient lived with her parents and two other siblings in a supportive environment. There were no reported psychosocial stressors, and the family was engaged in her healthcare needs. Despite her visual impairment, she demonstrated a positive attitude and actively participated in school activities, receiving assistance from teachers and peers.

During the antenatal period, the mother did not suffer from any significant infections that could have contributed to patients' condition. There was no maternal history of infections which impacts foetal development such as rubella, cytomegalovirus, or toxoplasmosis. The pregnancy was monitored regularly, and there were no complications. The eye examination findings of the patient were as shown in *Table 1*.

Table 1: Eye examination findings

Examination	Right eye	Left eye
External appearance	Normal eye size	Microphthalmos Noted
Visual Acuity (UCVA)	20/250	20/400
Visual Acuity (BCVA) Near BCVA	20/60 Not assessed	20/150 Not assessed
Refraction	+5.75/-0.50 × 112°	+4.50/-1.25 × 120°
Extra Ocular Muscle Movement	Free	Free
Intraocular Pressure (IOP)	14	15
Hirschberg's test	0^0	15^0
Anterior Segment	Normal	Normal
Vitreous	Clear	Clear
Retina	No detachment Choroidal thinning around the disc	No detachment Choroidal thinning with hyperpigmentation around the disc
Cup to Disc Ratio	Increased inferiorly	Increased inferiorly encroaching the fovea

Figure 1 is the fundus photograph of the patient's right eye. It shows an anomalous optic nerve with an increased cup diameter and deep cavitation inferiorly, along with thinning of the choroid around the optic disc. The upper part of the disc is well delineated, with significant peripapillary atrophy. A diagnosis of optic nerve coloboma was confirmed.

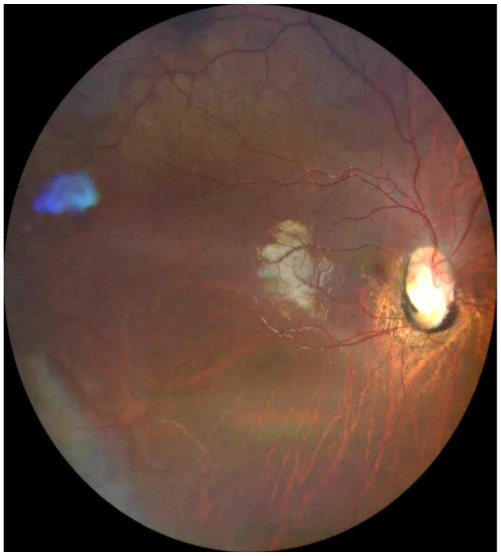

Figure 1: RE Optic Disc Coloboma (© David Munyi)

Figure 2 is the fundus photograph of her left eye. It shows a more significant optic disc coloboma compared to the right eye. There is a marked increase in cup diameter inferiorly, encroachment of the fovea by the coloboma, and a chorioretinal coloboma with hyperpigmentation central to the disc.

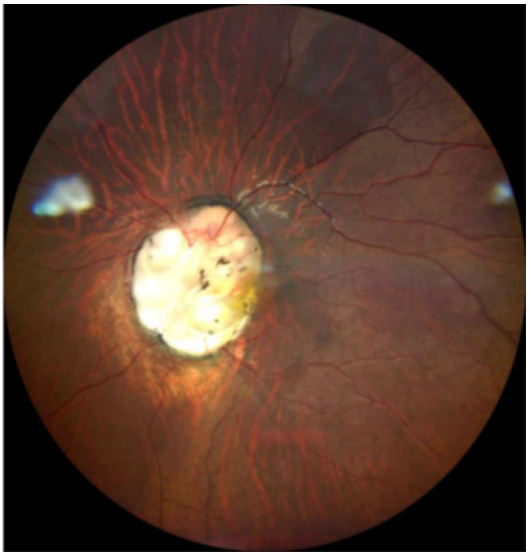

Figure 2: LE Optic Disc Coloboma (© David Munyi)

General examination did not reveal any of the other features associated with CHARGE Syndrome.

The diagnosis of optic nerve coloboma was made clinically based on the history and dilated fundoscopy findings. Genetic testing was not available.

Prognosis, follow up and Outcome.

The patient was discharged in stable condition and with spectacle correction. However, the primary limitation to visual acuity is the bilateral nature of the condition, with the left eye showing a more extensive coloboma and a poorer visual prognosis. She is expected to have a good prognosis for life since she did not have other manifestations associated with CHARGE syndrome.

Regular follow-up and documentation including a dilated fundus examination and fundus photography, to monitor for potential retinal detachment was recommended.

Discussion

Coloboma is a rare congenital ocular defect caused by the incomplete closure of the embryonic fissure during early development, leading to the absence of specific ocular tissues. It can affect both the anterior and posterior segments of the eye, with the optic disc, choroid, and retina being the most concerning sites in terms of visual prognosis(1). Posterior colobomas, particularly those involving the optic nerve, can significantly impair vision, especially when foveal involvement or retinal detachment occurs. Given the irreversible nature of optic disc coloboma, early diagnosis and consistent follow-up are crucial to preserving the remaining vision and preventing complications(2).

This case report highlights several important aspects of managing bilateral optic disc coloboma. First, the patient's presentation with bilateral involvement is particularly notable, as bilateral cases are associated with poorer visual outcomes compared to unilateral cases. In this instance, the larger and more extensive coloboma in the left eye (LE) resulted in more significant visual impairment, as evidenced by the discrepancy in visual acuity between the right eye (RE) and LE. The findings align with previous literature emphasizing that the extent of the coloboma, especially when bilateral, directly influences the prognosis(3).

The patient's diagnosis was supported by high-resolution imaging using the Zeiss Clarus 700 fundus camera. This technology allowed for detailed visualization of the optic disc and chorioretinal abnormalities, essential for both diagnosis and follow-up. While the imaging confirmed the absence of retinal detachment at the initial visit, the recommendation for serial fundus photographs every six months reflects good clinical practice. Monitoring for potential retinal detachment is critical, as timely intervention in such cases can preserve the remaining vision(4).

One key strength of this report is the comprehensive evaluation of the patient, including a thorough systemic examination to rule out CHARGE syndrome(5). Given that CHARGE syndrome is a known association in optic disc coloboma, it is essential to conduct a systemic workup to exclude life-threatening comorbidities. Although no systemic abnormalities were identified, the absence of genetic testing limits the diagnostic certainty, as it could have provided insights into possible hereditary factors or syndromic associations(6). This limitation reflects the challenges faced in resource-limited settings, where access to advanced diagnostic tools is often restricted.

In terms of management, the prescription of corrective spectacles was appropriate to optimize the patient's remaining vision. Regular follow-up will also play a vital role in monitoring for future complications, particularly retinal detachment, which remains a long-term risk in patients with posterior colobomas(7).

Psychosocial support is another important aspect highlighted by this case. Despite the patient's visual impairment, she has shown resilience, actively participating in school activities with the support of her family, teachers, and peers. This underscores the importance of a holistic approach to care, which not only addresses the medical aspects of the condition but also considers the psychosocial well-being of the patient(8).

Conclusion

This case report underscores the importance of early diagnosis, careful differentiation from similar conditions, and consistent follow-up in managing bilateral optic disc coloboma. While the absence of genetic testing and limited access to advanced care present challenges, the report provides valuable insights into managing complex ocular conditions in resource-limited settings. A policy which includes regular monitoring, prompt management of potential complications, and psychosocial support will be essential to ensuring the best possible outcome for this patient.

Ethical considerations

Informed Verbal consent was taken before the photos were taken. The name of the patient remained anonymous. No ethical approval has been provided but permission from Tenwek has been sort for publication.

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