

## A typical Location of a Lens Coloboma: A Case Report of 2 Patients

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### Abstract

### Case Report

Lens coloboma is a rare congenital anomaly caused by abnormal zonular development. We report two cases: a 39-year-old woman with left-eye lens coloboma associated with nuclear cataract, and a 3-year-old child with right-eye lens coloboma associated with congenital glaucoma and high myopia. The condition results from localized absence of zonules leading to lens deformation and may be associated with other ocular abnormalities. Early diagnosis and follow-up are essential to preserve visual function.

**Keywords:** Lenticular Coloboma, Zonular Deficiency, Atypical Location.

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## INTRODUCTION

Colobomas are congenital malformations of the eye, caused by incomplete closure of the foetal cleft, and all structures of the eye can be affected. However, lens coloboma remains an extremely rare site of this malformation, and our case is one of only a handful described in the literature. As with all forms of coloboma, lens coloboma may be associated with ocular malformations such as cataracts, microphthalmia or lens ectopia, the latter being one of the differential diagnoses of lens coloboma [1, 2].

### Case 1

The case of a 39-year-old female patient with no particular pathological history is presented. She presented to the ophthalmology consultation with a progressive decline in visual acuity that had been evolving for one year. Examination of the left eye revealed corrected visual acuity of 20/63 and automatic refraction of -1.50 (-4.25 at 156°). A thorough examination of the anterior segment of the eye revealed a supra-temporal crystalline coloboma, accompanied by a zonular defect ranging from 11h to 2h, and the presence of a nuclear cataract [Figure 1,2]. A subsequent examination of the iris and fundus was conducted, but no significant findings were revealed. A thorough examination of the right eye did not reveal any notable abnormalities.

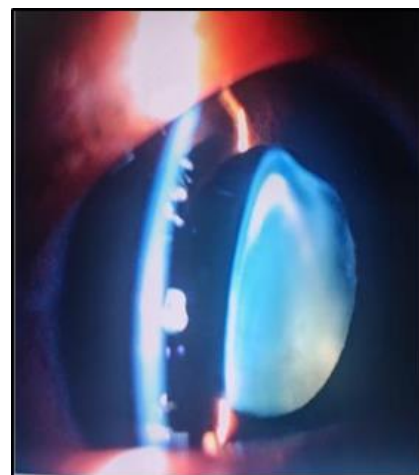


Figure 1

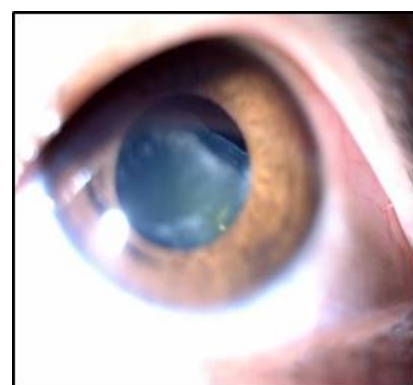
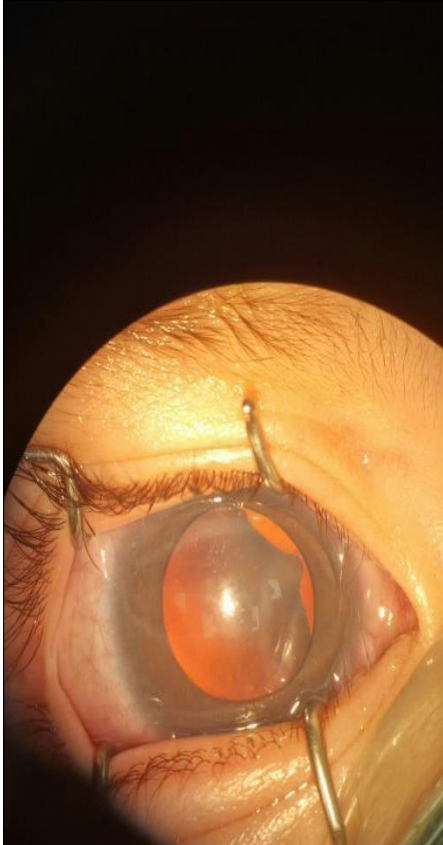


Figure 2

## Case 2

We report the case of a 3 year old child, followed in our department for unilateral congenital glaucoma of the right eye. Examination of the right eye with sedation revealed an automatic refraction of -11.50 (-2.25 at 156°). Examination of the anterior segment revealed a supra-nasal crystalline coloboma with a zonular defect ranging from 1h to 4h (Figure 3). Examination of the iris revealed an iridectomy at 11h and the fundus revealed a papillary excavation c/d:6/10. A thorough examination of the left eye did not reveal any notable abnormalities.



**Figure 3**

## DISCUSSION

Lens coloboma is defined as an incomplete formation of the lens due to incomplete closure of the foetal fissure, which is associated with an absence of zonules, producing an indented appearance of the periphery of the lens, often inferonasal. Colobomas of the lens are categorised into two distinct types: typical colobomas, which manifest at the level of the embryonic fissure, and atypical colobomas, the pathophysiology of which remains uncertain and does not occur at the level of the embryonic fissure [1, 2].

The embryological origin of the atypical coloboma remains uncertain; various theories suggest that it could result from rotation of the foetal fissure or develop following an inflammatory process. It could also result from the persistence of mesodermal tissue of embryonic vascular origin blocking the anterior growth of the neuroectoderm, thus causing a defect in the ciliary body and iris [2].

The term "lens coloboma" is often a misnomer and might be more appropriately referred to as "zonule" or "ciliary body coloboma," as the condition actually involves the segmental notching and contraction of the lens due to the absence of zonules at this site [3].

The failure of zonules to form in the anterior secondary vitreous or in the marginal bundle of Druault during the third to fourth month of gestation can be attributed to either the failure of condensation and differentiation of the vitreous substance, or the failure of the lens to induce such changes [4].

## CONCLUSION

Lens coloboma, a rare malformation, can lead to severe ocular complications, emphasising the importance of early detection and appropriate follow-up to preserve visual function in affected patients.

The authors declare that they have no link of interest concerning this article.

The authors declare that they obtained a written informed consent from the patients and/or volunteers included in the article and that this report does not contain any personal information that could lead to their identification.

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