

## Multiple Types of Coloboma in an Otherwise Healthy Patient: A Case Report

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### ABSTRACT

**Purpose:** this study aimed to report a case of a unilateral lenticular coloboma super imposed over bilateral iris and optic nerve coloboma in a healthy patient, colobomas are rare congenital malformation of the neuroectodermal tissue of the optic cup. **Materials and Methods:** this case of medically free patient who complained of poor vision since early childhood that has progressed in the last five years. Patient underwent thorough and complete ophthalmic investigation. **Results:** fundus photography revealed inferior optic nerve colobomas in both eyes and no uveal colobomas. On slit lamp examination, bilateral inferior iris coloboma were noted. Systemic examination was unremarkable. Patient was otherwise healthy. **Conclusion:** our report showed the need of more studies to be done in order to have a better understanding of the prevalence, management and genetics related to this condition in our population.

**Keywords:** optic nerve coloboma, iris coloboma, optic nerve malformation, optic nerve anomaly.

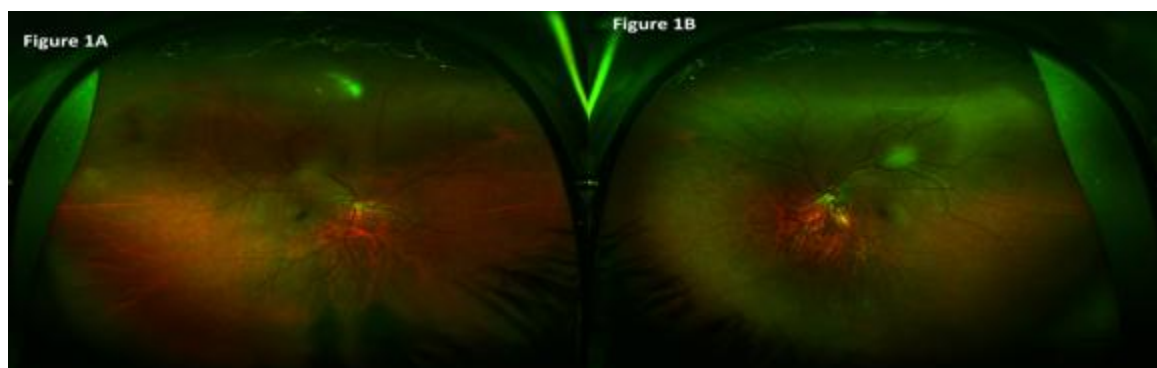
### INTRODUCTION

Colobomas are a rare congenital malformation of the neuroectodermal tissue of the optic cup. Typically, during the 7<sup>th</sup> week of gestation, the optic cup is formed by the closure of the choroidal fissure, leaving a round opening that becomes the pupil. Failure of this process results in a persistent cleft that can involve the anterior or posterior parts of the eyeball <sup>(1)</sup>. Coloboma iridis – where the malformation occurs anteriorly and is limited to the iris - is the most common form. Anterior colobomas may also involve the ciliary body. Posterior colobomas may involve the optic nerve, retina and choroid <sup>(2)</sup>. It is estimated that the incidence of coloboma is around 0.7 cases per 10,000 live births. Although colobomas are not usually familial, they have been observed to be occasionally inherited in an autosomal recessive or, rarely, an X-linked pattern. Colobomas may also be associated with a with PAX2 gene mutations <sup>(2,3)</sup>.

### CASE

Our case was a 36-year-old medically free male referred to our hospital from another city for

medical evaluation. The patient complained of poor vision since early childhood that has progressed in the last five years. The patient was suffering from occasional photophobia. Family ocular history was positive: The patient's brother was suffered from decreased vision since childhood as well he did not seek any medical advice. Distant unaided visual acuity (VA) was 6/18, 6/60, and best-corrected visual acuity (BCVA) was 6/12 and 6/21 for the right and left eyes respectively. The patient had no nystagmus or strabismus and intraocular pressure (IOP) was normal in both eyes. On slit lamp examination (SLE), the eyelids, conjunctiva and cornea were normal bilaterally. The anterior chambers of both eyes were deep and quiet. A bilateral inferior iris coloboma was noted. Additionally, developmental cataracts that are not visually significant were noted in both eyes, as well as an inferior lenticular coloboma in the left eye. On fundus examination, there were inferior optic nerve colobomas in both eyes and no uveal colobomas (**Fig.1 A,B**). Systemic examination showed no abnormalities or apparent congenital anomalies.



**Figure 1A, B: right and left eye, respectively, showing inferior optic nerve coloboma.**

## DISCUSSION

An extensive search of literature shows no case reports of isolated colobomas from Saudi Arabia; however, it has been reported in the context of syndromes such as Kabuki syndrome and C12orf57 mutation related colobomatous microphthalmia <sup>(4,5)</sup>. The incidence and prevalence are not available for this region; more studies are needed to be done. This data are interesting because of the high prevalence of consanguinity among the Saudi population. Consanguinity means that diseases with autosomal recessive patterns of inheritance are more common.

The exact etiology of coloboma is still being explored. **Warburg** has noted that colobomas, microphthalmia, and anophthalmia may be pathophysiologically related at the molecular genetic level <sup>(3)</sup>. Coloboma is routinely found as part of a syndrome, the most common of these being the charge syndrome (Coloboma, heart problems, choanal atresia, retardation of growth and development, genitourinary abnormality). **Asai-Coakwell et al.** implicated deletions involving Growth Differentiation Factor 6 (GDF6) in coloboma, microphthalmia and other ocular anomalies <sup>(6)</sup>. PAX6, although was identified to play a fundamental role in ocular development, does not appear to be a primary cause for colobomas in humans <sup>(7)</sup>.

Our case is unique because the iris and optic nerves were involved bilaterally, while the lens was involved only in the left eye. True colobomas of the lens are exceedingly rare – the lens arises from a different embryological origin than the rest of the optic cup. Typically, chorioretinal colobomas involving the ciliary body and zonules distort and flatten the lens. This flattening leads to lenticular astigmatism that can adversely affect visual acuity. Conditions such as Marfan syndrome, ADAMTSL4 mutation, and lens subluxation may cause similar lens malformations <sup>(7)</sup>.

Treatment and diagnosis of coloboma represented a challenge in all affected patients. Coloboma treatment largely depends on the context in which it presents and the degree to which visual acuity is affected. Refractive error is treated with corrective lens and intra ocular lenses if needed. A valid concern patients may present with cosmesis, particularly in lesions involving the iris with heterochromia and pupil displacement. Colored contact lenses or surgical correction can be considered <sup>(7)</sup>.

Colobomas also predispose patients to complications. Cataracts tend to form at a younger age in coloboma patients. Zonule malformations and the flattened lens shape complicates cataract surgery. Patients with posterior colobomas are more susceptible to retinal detachment. These complications may compromise the vision and require interval monitoring every 6-12 months <sup>(7)</sup>.

## CONCLUSION

We have presented an atypical presentation for coloboma, the first of its kind in Saudi Arabia. In our case, we have obtained the patient's medical history with a complete ophthalmic and physical examination. Manifestations and complications of coloboma were investigated and came back negative. Corrective lenses were prescribed to treat the decrease in vision. Regular follow up is scheduled to monitor the patient for the development of cataract, neovascularization or retinal detachment. However, more studies need to be done in order to have a better understanding of the prevalence, management and genetics related to this condition in our population.

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