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Case Report

Bilateral Iris and Chorioretinal Coloboma: A Case Report

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Abstract

Coloboma is a congenital anomaly characterized by a defect in one or more structures of the eye. It results from a failure in the closure of the colobomatous fissure, which typically occurs between the 5th and 7th week of embryonic development. We present the case of a 19-year-old patient from a non-consanguineous marriage, with no notable pathological history, who consulted for a progressive decrease in bilateral visual acuity. Examination with a slit lamp and fund us copy revealed bilateral iris and chorioretinal coloboma in both eyes. A thorough physical examination was performed to rule out other associated mal formations, and it returned normal. Regular ophthalmological monitoring is necessary to detect any risk of retinal detachment, the main complication associated with coloboma. It is also essential to evaluate the possibility of systemic involvement, which can have serious implications for overall prognosis.

Keywords: Iris coloboma; Chorioretinal coloboma; Malformation; Eye

Introduction

Coloboma is a developmental anomaly of the eye that occurs during embryonic development, typically between the 5th and 7th weeks of gestation. This defect results from the failure of the fetal fissure to close and manifests as gaps in the inferonasal quadrant of the eye, where tissue is missing. Ocular coloboma is often associated with systemic anomalies when caused by chromosomal abnormalities. It can affect various structures of the eye, leading to gaps in the iris, choroid, retina, lens, eyelid, or optic nerve. Depending on the location of these gaps, several types of colobomas are distinguished [1]. Coloboma can affect one eye or both eyes simultaneously. The clinical diagnosis of chorioretinal coloboma is generally straight forward when accompanied by involvement of the anterior segment of the eye [2].

Case Presentation

This is a case of a 19-year-old female patient from a non-consanguineous marriage, with no notable pathological history, who presented to our clinic with a progressive decrease in bilateral visual acuity over the past 2 years. On clinical examination, visual acuity was 4/10 in the right eye (OD) and 3/10 in the left eye (OG). Examination of the anterior segment revealed a deformed pupil in both eyes, with an inferior nasal notch causing a loss of its round appearance (Figure 1). Fundus examination showed an inferonasal chorioretinal coloboma in the right eye, sparing the optic disc, characterized by a well-defined, whitish area with few blood vessels. In the left eye, there was a papillary coloboma extending in to the inter papillary-macular and nasal regions (Figures 2 and 3). In light of these findings, a thorough physical examination was performed to rule out other associated malformations (cardiac, genitourinary, ENT, etc.), and the results were normal.

Discussion

Congenital ocular colobomas result from a failure of closure of the fetal fissure during organogenesis [1]. The prevalence of colobomas

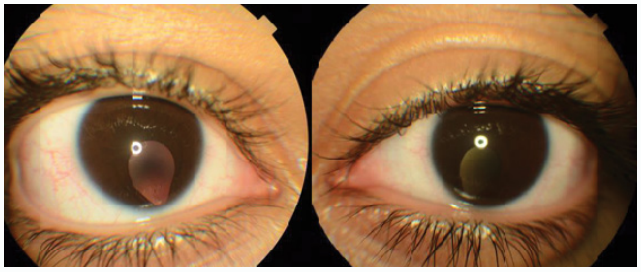

Figure 1: Inferonasal iris coloboma in both eyes.

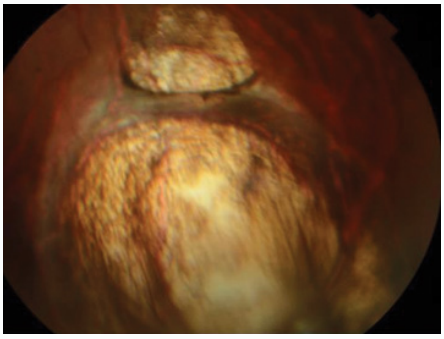

Figure 2: Fundus appearance of the right eye showing an inferonasal chorioretinal colobomas paring the optic disc.

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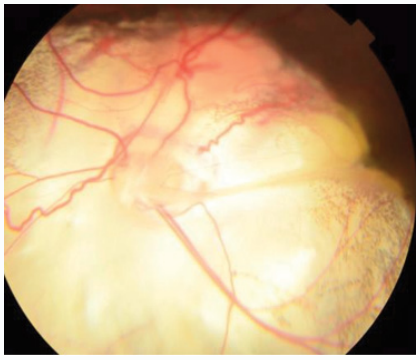

Figure 3: Fundus appearance of the left eye showing a papillary coloboma extending in to the inter papillary-macular and nasal regions.

is 0.5 to 0.7 per 10,000 births [2]. During embryonic development, the eye naturally features a fissure known as the colobomatous fissure, typically located in the lower and slightly nasal position, extending from the optic nerve to the papillary margin. This fissure should normally close once eye development is complete. However, incomplete closure of this fissure can lead to partial or complete absence of certain eye structures, resulting in colobomas [3]. These anomalies can affect one or both eyes. Colobomas may arise from genetic mutations or exposure to toxic environmental factors. They are a significant cause of visual impairment and childhood blindness. Congenital ocular colobomas are frequently observed in various multi systemic syndromes, often accompanied by neurological, craniofacial, or other systemic developmental defects. Additionally, acquired colobomas can also form as a result of traumatic injury or following ocular surgical procedures [1,4]. The clinical diagnosis of chorioretinal coloboma is generally straight forward when accompanied by involvement of the anterior segment of the eye. However, it can be challenging to establish when it is isolated, especially in young children. In such cases, the presence of a whitish lesion in the fundus may suggest various retinal pathologies, including tumor-related, infectious, or genetic causes, among others [5].

Management of coloboma includes two main aspects:

Ophthalmological Care: For iris coloboma, the use of colored (cosmetic) or corrective contact lenses may be recommended to reduce are. In cases of chorioretinal coloboma with associated retinal detachment, surgical intervention might be necessary, though it is complex due to the delicate nature of the eye. Regular ophthalmological monitoring is crucial to detect complications, especially retinal detachment, which is a major concern [6].

Systemic Evaluation: While most coloboma cases are idiopathic and sporadic, various modes of inheritance have been described, sometimes associated with different syndromes such as charge syndrome, Meckel-Gruber syndrome, Jorgen-Larson syndrome, Lenz syndrome, and Catel-Manzke syndrome, among others. Therefore, a comprehensive physical examination is essential to identify any associated malformations or neurological disorders and to assess for multi systemic involvement [7]. In outpatient's case, the physical examination revealed no additional abnormalities.

Conclusion

Coloboma is a congenital malformation that can affect various ocular structures influencing visual prognosis. The standard management for any coloboma primarily involves regular clinical and para clinical monitoring. This approach allows for the early detection and prompt treatment of potential complications.

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