Case Report

Lens Coloboma and Associated Ocular Malformations

Juanjuan Li, Xuan Ma, Zhulin Hu
Department of Ophthalmology, Yunnan Red Cross Hospital, Kunming 650021, China

Abstract

Purpose: Lens coloboma is a rare congenital disorder of crystalline lens characterized by notching of the equator of the lens. Coloboma can occur in isolation or with other ocular malformations. The authors reported 5 cases of lens coloboma associated with some ocular malformations.

Methods: Case reports. The clinical features, imaging findings and associated ocular malformations were reported.

Results: One case was not associated with ocular anomalies. One patient was involved with iris and choroid coloboma; another subject suffered from total cataract; and the fourth patient was affected by lens dislocation. The remaining case included aniridia. The pathogenic mechanisms and relationship of the ocular malformations have been discussed.

Conclusion: Lens coloboma can be associated with other ocular malformations, such as iris coloboma, choroid colobma, localized or total caratact. (***Eye Science 2011;26:109-111***)

Keywords: Lens coloboma; iris coloboma; choroid coloboma; caratact

Introduction

Coloboma of the lens is often characterized as invagination of the equator of the lens. The most frequently seen form is the inferior-nasal location. Coloboma usually occurs unilaterally, and if bilaterally, is symmetrical coloboma. The zonules may be absent in the coloboma regions. Lens coloboma can be associated with other ocular malformations, such as iris coloboma, choroid colobma, localized or total caratact.

Case reports

Case 1: A 11-year-old boy presented with bilateral decreases in vision acuity (VA). There was no positive family history or any systemic disease. The best corrected VA was 20/20 with-4.00 diopter correction in both eyes. Iris appearance, pupils were not obvious. After papillary dialation, slit-lamp biomicroscopy revealed inferior notching of the crystalline lens in the right eye, zonular was absent with corresponding lens coloboma (Figure 1). Fundus examination showed no retinochoroidal coloboma. The patient was diagnosed with bilateral lenticular coloboma.

Case 2: A 17-year-old man was referred to our department with poor VA in the left eye since his childhood. The patient had VA of one meter counting fingers and VA improved to 20/200 with -1.00 diopter. Ophthalmologic examination revealed a small coloboma situated in the supero-nasal coloboma and total iris coloboma (Figure 2). Fundus examination showed normal outcomes. The patient was diagnosed with aniridia lenticoular colobma.

Case 3: A 8-year-old boy presented with bilateral decreases in VA. Uncorrected VA was 1/20 for both eyes. The best corrected VA was 10/200 in both eyes with-2.50 diopter (D) correction. Ophthalmologic examination revealed bilateral inferior iris and lens coloboma (Figure 3A). Intraocular pressure was 14 mm Hg bilaterally. Funduscopic examination revealed choroid coloboma without retinal detachment (Figure 3B). The subject was diagnosed with bilateral congenital iris, lens and choroid coloboma.

Case 4: A 4-month-old girl was referred due to white pupil in her left eye. The pupils were briskly reactive to light and had lens coloboma in inferior-nasal locations with deficient zonules in the notched part of the lens. Total cataract was also observed (Figure 4). Ophthalmologic examination indicated that the right eye was normal. The girl was diagnosed with congenital lens coloboma and cataract.

Case 5: A 36-year-old man was admitted to our

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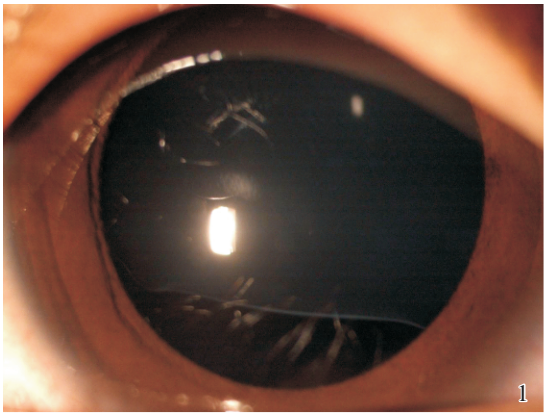
Corresponding author: Juanjuan Li, Department of Ophthalmology, Red Cross Hospital, No.176, Qingnian Rd., Kunming, China, E-mail: ljj800502@163.com



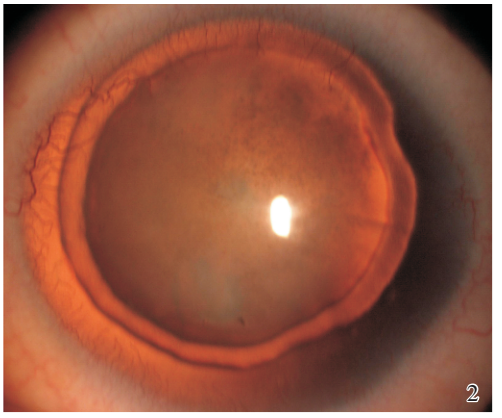
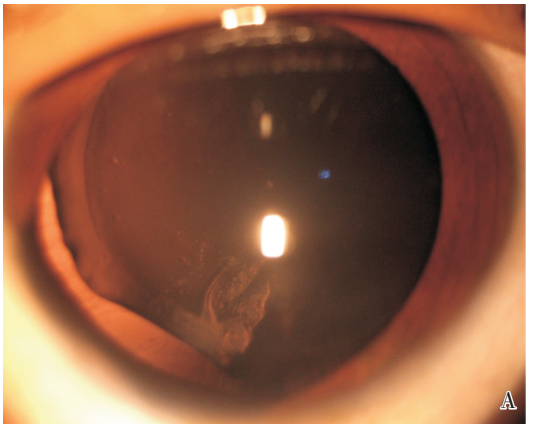


Figure.1 Slit lamp photograph showed dilated right eye showing notching of the lens equator inferiorly with absence of zonules in the corresponding area in Case 1.

Figure.2 Slit lamp photograph revealed a small coloboma situated in the supero-nasal coloboma and total iris coloboma in Case 2.



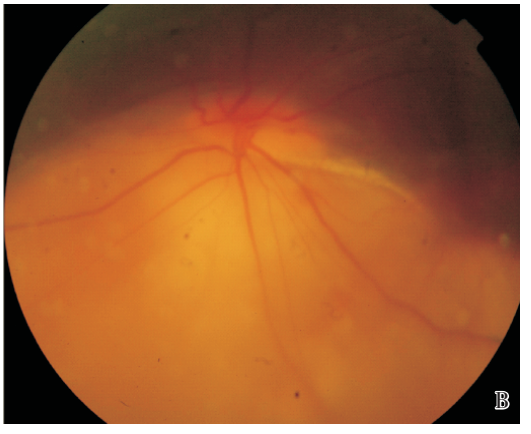
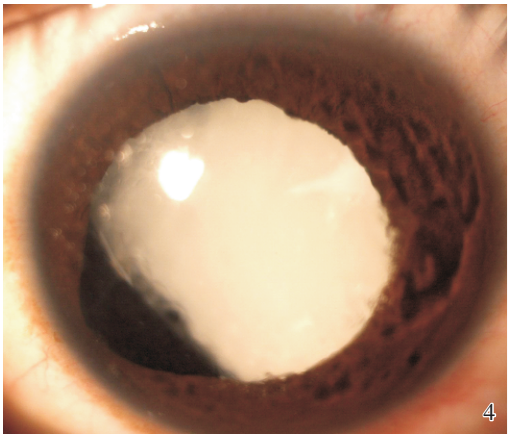


Figure.3 **A.** Slit lamp photograph revealed retroillumination showing inferior iris and lens coloboma in Case 3. **B.** Fundus photograph revealed choroid coloboma without retinal detachment in Case 3.



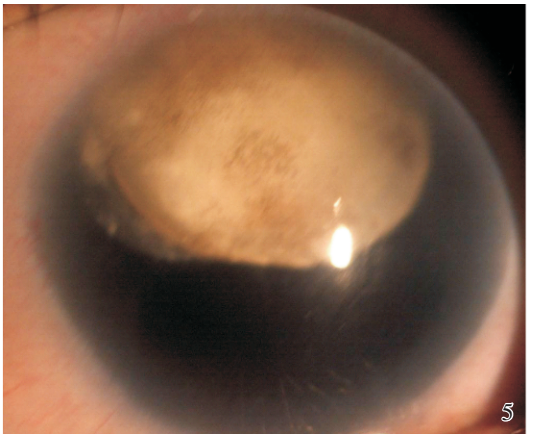


Figure.4 Slit lamp photograph revealed lens coloboma in inferior-nasal locations with deficient zonules in the notched part of the lens, and total cataract was seen in Case 4.

Figure.5 Slit lamp photograph revealed inferior lens were absent with a corresponding zonular coloboma. The lens were opaque totally and dislocated upwardly in Case 5.

clinic with complaints of low VA in both eyes since his childhood. Birth and family history provided no remarkable clues. VA was counting fingers in both eyes and failed to be improved after correction. Both

eyes had poor fixation. Biomicroscopic examination revealed inferior lens were abscent with corresponding zonular coloboma. The lens were opaque totally and dislocated upwardly (Figure 5). No associated retinochoroidal coloboma was noted. Despite the pendular nystagmus, his eyes movement was normal in all directions.

Discussion

Lens coloboma may typically arise in inferonasal location, and also occur in other locations. Toxic, inflammatory or genetic factors, etc may be considered to be related to the weakness of zonular fibers in the equatorial area of the lens[1]. Ciliary body or iris cyst, persistent fetal vasculature vessels may disrupte zonular and lenticular development[2]. Faulty development or deficiency of the zonules in the equator results in the absence of pull force exerted by the zonular fibers.

Lens coloboma can also be associated with other ocular malformations, such as iris, ciliary and choroidal coloboma, possibly due to faulty closure of the fetal tissue during the fourth to fifth weeks of gestation[3]. Congenital anirida with lens coloboma is considered as the result of mutation of PAX6 gene. PAX6 genes are the family of developmental control genes that encode nuclear transcription factors, which also regulates the organogenesis of the eye[4]. Lens coloboma is often associated with lertain form of cataract, either posterior cortical, nuclear or total cataract[5]. Localized or total cataract was also observed in our cases. Except for cataract, lens coloboma is potentially associated with other lenticular anomalies, such as ectopia lentis, lens opacities and subluxation of the lens.

During review of the literature, lenticular coloboma accompanied by snowflake retinal degeneration[5], retinal detachment[6], optic disc coloboma[7], persistent papillary membrane[8], and optic nerve hypoplasia with orbital hemangioma[9] has been reported.

In conclusion, lens coloboma cases should be further evaluated for multiple potential malformations.

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