

Bilateral congenital uveal coloboma concurrent with retinal detachment

Zi-Jing Huang, Jing-Sheng Yi, Wei-Qi Chen

Joint Shantou International Eye Center of Shantou University and the Chinese University of Hong Kong, Shantou 515041, Guangdong Province, China

Correspondence to: Wei-Qi Chen. Joint Shantou International Eye Center of Shantou University and the Chinese University of Hong Kong, 69 North Dongxia Rd, Shantou 515041, Guangdong Province, China. cwq@jsiec.org

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Dear Editor,

We are writing to present a bilateral uveal coloboma concurrent with retinal detachment (RD) in a 16-year-old girl. Colobomas are rare, congenital and potentially blinding malformations caused by incomplete closure of embryonic optic fissure^[1]. Uveal colobomas typically involve the inferior part of the choroid and/or iris and may sometimes cause retinal detachment^[2]. The visual prognosis of uveal colobomas is highly variable, which depends on its size and location. Poor visual acuity is often present when the uveal coloboma involves the macula. In this study, we report a case of a young patient with bilateral iridochoroidal coloboma who suffered from macula-involved RD in both eyes, presenting the clinical characteristics and management of uveal colobomas-associated RD.

Ethical Approval This study complied with the tenets of the Declaration of Helsinki. Written informed consent was obtained from the patient's mother for publication of this case report and any accompanying images.

Case Report A 16-year-old girl presented with progressively bilateral painless visual loss for 2wk. She reported poor vision since early childhood without remarkable medical and family history except laser treatment in her left eye several months ago. On examination, her best corrected visual acuity (BCVA) was 20/400 on both eyes and intraocular pressure was normal. Cycloplegic refraction revealed a compound myopic astigmatism of -4.75 DS/-2.00 DC in the right eye and -3.75 DS/-1.25 DC in the left eye. Biological measurements

demonstrated an axial length of 25.06 and 25.19 mm in her right and left eyes, respectively. Nystagmus in both eyes was noted, especially when one of the eyes was covered. Slit-lamp examination showed a black notch at the inferior edge of the pupil suggestive of iridal coloboma (Figure 1). The cornea and lens in both eyes were normal. Fundus examination revealed large and oval gray-white lesions involving the macula and the inferior retinal quadrant in both eyes (Figures 2A and 3A). Bilaterally symmetrical choroidal coloboma concurrent with RD was further indicated by B-scan. Of note, detachment of the retina was found both inside and outside the colobomatous area (Figures 2B and 3B). Optical coherence tomography revealed macula-involved RD with regional retinoschisis, retina thinning, loss of ellipsoid zone, and an unidentified macular fovea (Figures 2C and 3C). A diagnosis of bilateral iridochoroidal coloboma consensus with RD was made.

The girl underwent 23-gauge pars plana vitrectomy (PPV) following silicone oil tamponade in both eyes successively. During surgery, detachment of the retina was found extending from the inside to the superior and temporal area outside the coloboma. Retinal tears were present within the colobomatous area on both eyes, which were treated with cryotherapy. Laser photocoagulation was applied at the margin of the coloboma. Her left eye showed successful retinal reattachment using dilated fundus examination and optical coherence tomography (Figure 2D and 2E), with the BCVA fluctuating between 20/100 and 20/80 during the follow-up periods. Silicone oil removal was performed 10mo after the primary procedures. Retinal thinning was found during the 1-year follow-up with a final BCVA of 20/100. In her right eye, the retina was found nearly reattached with a small gap over the colobomatous area at 1wk after surgery (Figure 3D). One month later, complete reattachment of the retina was observed (Figure 3E) and BCVA improved to 20/200. She had silicone oil removed several months later. At the 1-year follow-up after silicone oil removal, the retina remained completely attached and BCVA remained stable at 20/200. It was noted that the girl developed cortical cataract in both eyes several months after the primary surgery. Since the cataract was mild and showed no significant progression until the last follow-up, an observation was still conducted instead of cataract surgeries.

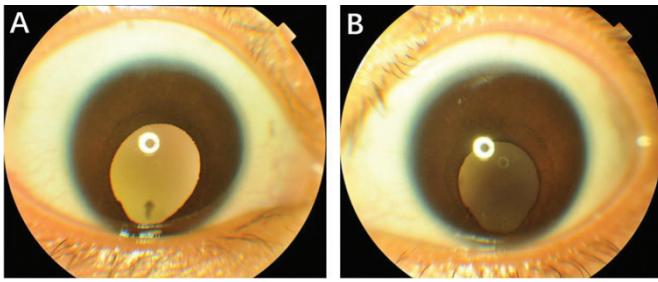


Figure 1 Anterior segment photography showed coloboma of the iris presenting with a black notch at the inferior edge of the pupil A: Right eye; B: Left eye.

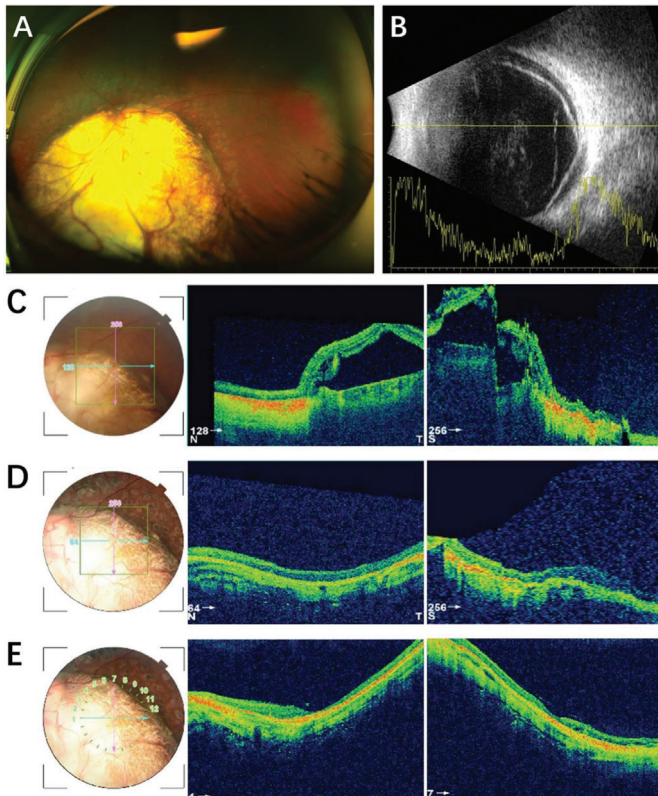


Figure 2 Ocular examination of the left eye A: Scanning laser ophthalmoscopy revealed a large and oval gray-white lesion in inferior macula with laser spots around the lesion before surgery; B: B-scan indicated the presence of choroidal coloboma consensus with retinal detachment; C: Detachment of the retina at the margin of the coloboma before surgery; D: Well reattached retina at 1wk after surgery; E: Complete retinal attachment with retinal thinning at the 1-year follow-up; C-E: Images of optical coherence tomography.

Uveal colobomas are congenital and potentially blinding disorders with a prevalence of about 0.5-2.6 per 10 000 people and contributes to up to 10% of childhood blindness^[3]. Uveal colobomas are often associated with microcornea and microphthalmia as well as other ocular disorders, such as cataract, nystagmus and amblyopia, myopia, glaucoma, and RD^[4]. Patients with choroidal colobomas present increased risk of RD, with an incidence rate ranging from 2.4%-42% as reported^[5-7]. In this study, we present a case of bilateral

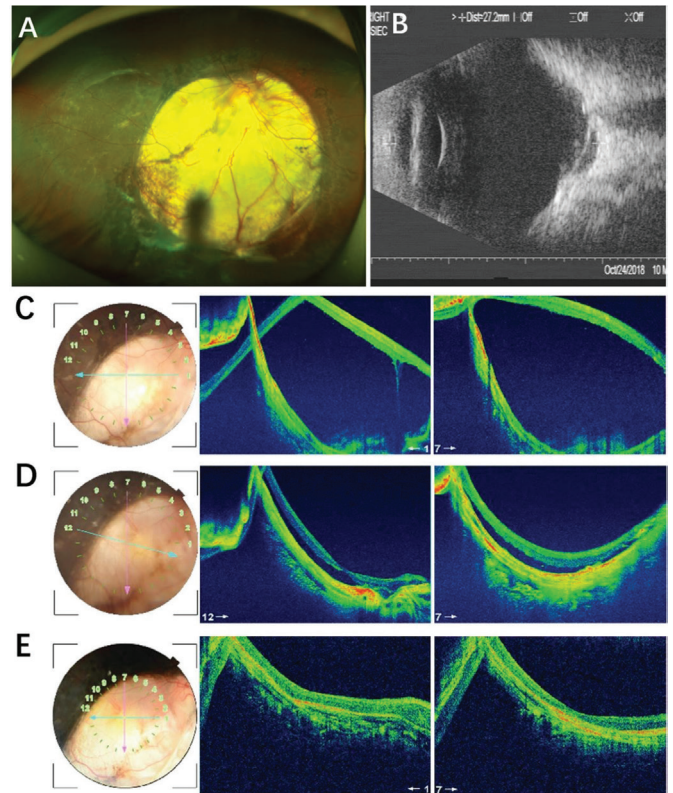


Figure 3 Ocular examination of the right eye A: Scanning laser ophthalmoscopy revealed a large and oval gray-white lesion in inferior macula before surgery; B: B-scan indicated choroidal coloboma and retinal detachment; C: Detachment of the retina inside the colobomatous area before surgery; D: Almost reattached retina with a small gap over the colobomatous area at 1wk after surgery; E: Complete retinal attachment at the 1-month follow-up. C-E: Images of optical coherence tomography.

iridochoroidal coloboma-related RD who had successful repair after long-term follow-up, which is rarely reported in the literatures.

Uveal coloboma-associated RD often requires surgical intervention to reattach the retina and improve visual acuity. Scleral buckling has been proposed as a treatment strategy when peripheral retinal tear rather than tears in the colobomatous zone is evident, with an anatomical success rate of 57%^[8]. However, the tears causing RD in the colobomatous eyes are often hidden within or at the edge of the colobomatous lesion, making them difficult to identify^[9]. Therefore, PPV combined with silicone oil or gas is highly recommended as it provides guarantee for identification and management of potential retinal tears. In this case, we carried out a thorough dilated fundoscopic examination and found no visible tears in both eyes. Given this situation, we performed PPV combined with laser photocoagulation around the colobomatous area, transcleral cryotherapy inside the colobomatous area, and subsequent silicone oil endotamponade. Due to the absence of choroidal pigment of the coloboma, laser treatment always

fails to seal retinal breaks inside this area. We therefore conducted transcleral cryotherapy for these breaks, which was proved to be effective.

The primary reattachment rate using PPV and silicone oil tamponade was once reported to be 81.7%^[10]. However, it was found that a considerable proportion of patients still had their silicone oil remaining for various reasons^[10-13]. In this level, they could not be defined as an eventual success since it is unclear whether retina could be attached after silicone oil removal, particularly in colobomatous eyes. Our patient had silicone oil removed in both eyes and presented with completely attached retinas in the 1-year follow-up, which we believed, could be demonstrated as a definite success.

Uveal colobomatous eyes with RD often present poor visual prognosis particularly when the macula is involved^[14]. One research reported that 60% of patients had the final BCVA of hand movement or worse^[11], whereas another study demonstrated nearly 70% of patients had an average BCVA of 10/200 at 13mo after surgery^[10]. A review of pediatric patients with colobomas found that 38% of cases undergoing RD repair present an equal or better BCVA compared with non-surgical eyes^[15], indicating the necessity and effectiveness of surgery. In this study, better BCVA was achieved in both eyes after surgery (20/400 to 20/200 OD and 20/400 to 20/100 OS) and maintained rather stable until the 1-year follow-up although mild cataract was present. Our case presents a satisfactory visual outcome as compared to previous studies, possibly due to incomplete involvement of the macula so that paracentral fixation is still available.

Uveal colobomas can exist either in isolation or associated with systemic abnormalities^[16-17]. Our patient did not present systemic abnormalities including cardiac and hearing disorders, mental retardation, and denied any family history. Nevertheless, it is highly recommended that ophthalmologists prompt a careful family history and systemic examination including ultrasonic cardiogram, audiometric tests, kidney ultrasound, spine imaging, *etc.* in patients with uveal colobomas.

In conclusion, we report a young girl with bilateral uveal coloboma consensus with rhegmatogenous RD. PPV combined with laser photocoagulation around the colobomatous area, transcleral cryotherapy inside the colobomatous area, and silicone oil tamponade, could be effective alternatives for such complex RD. In addition, ophthalmologists should remain careful and thorough in assessing the patients with uveal coloboma to avoid any delay in the diagnosis and management of systemic disorders.

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Conflicts of Interest: Huang ZJ, None; Yi JS, None; Chen WQ, None.

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