• Letter to the Editor •

Lens coloboma with bilateral ectopia lentis in Marfan syndrome: a case report

Zhen–Bo Zhao[1,2], Kai–Li Tang[1,2], Yu–Xi Ding[1], Ji–Kun Yang[1,2], Huan–Ping Wang[1], Li–Wei Ma[1,2]

[1]Aier Eye Medical Center of Anhui Medical University, Hefei 230031, Anhui Province, China
[2]Aier Excellence Eye Hospital, Shenyang 110000, Liaoning Province, China
Correspondence to: Li-Wei Ma. Aier Eye Medical Center of Anhui Medical University, Hefei 230031, Anhui Province, China. maliwei@aierchina.com
Received: 2023-11-22 Accepted: 2024-09-17

DOI:10.18240/ijo.2025.02.21

Citation: Zhao ZB, Tang KL, Ding YX, Yang JK, Wang HP, Ma LW. Lens coloboma with bilateral ectopia lentis in Marfan syndrome: a case report. *Int J Ophthalmol* 2025;18(2):358-361

Dear Editor,

We report a rare case of lens coloboma and several notches with bilateral ectopia lentis in Marfan syndrome. This case was approved by the Ethics Committee of Aier Excellence Eye Hospital (2023KJB0004). Written informed consent was obtained from the patient. Lens coloboma and Marfan syndrome are of low incidence and seldom observed in the clinic. A few cases of lens coloboma with a slight notch have been reported[1-2], but lens coloboma and several notches with bilateral ectopia lentis in Marfan syndrome have rarely been reported. Marfan syndrome is frequently linked with ectopia lentis but is rarely associated with congenital lens abnormalities. Through this case report and literature study, we can gain a deep understanding of this type of disease, which is convenient for comprehensive diagnosis and treatment in the future.

CASE REPORT

A 33-year-old female visited our clinic, complaining of decreased vision in both eyes (worse in left eye) for 3mo. Marfan syndrome was passed down through three generations of the family (mother, our patient and her brother, and nephew). Physical examination revealed a height of 180 cm, a weight of 65 kg, long and thin extremities, spider-like fingers, and cardiovascular abnormalities (Figure 1).

Ocular examination: Uncorrected distance vision acuity was 20/40 in the right eye and 20/400 in the left eye, and corrected distance vision acuity was 20/25 and 20/63 respectively. Intraocular pressures were 16.5 and 18.5 mm Hg. On slit lamp examination, no conjunctival congestion, the anterior chamber depth was uneven, and the pupil could not be fully dilated in both eyes. After mydriasis, superionasal dislocated lens was detected in both eyes, along with lens coloboma (extending from 4 to 12 o'clock in right eye, 2 to 8 o'clock in left eye) and stretched zonules as well. The lenses of both eyes were clear along with several notches and the absence of zonules at the equator (Figure 2). There was no obvious abnormality in either fundus.

Corneal topography (OPD-Scan III) revealed regular corneal curvatures [right (R)=42.38 D, left (L)=43.11 D]. The result of IOL-Master700 showed that the axial length was shorter (R=21.13 mm, L=22.20 mm), the anterior chamber depth was shallow (R=2.48 mm, L=1.74 mm), and the lens thickness were normal (R=4.32 mm, L=4.45 mm). Despite the relatively normal biometry of cornea and axial length, auto refraction/keratometer (NIDEK ARK-1) indicated high astigmatism and myopia [R=-0.5 degree of spherical (DS)/-5.25 degree of cylindrical (DC)×112, L=-8.75 DS/-11.0 DC×4], the myopia and astigmatism were due to the lens abnormalities. Ultrasound biomicroscopy revealed that anterior chamber depth was uneven, lens subluxation, the ciliary body became thin (ciliary body thickness: R=0.98 mm, L=0.92 mm), and the ciliary process was not clear (ciliary process length: R=0.71 mm, L=0.62 mm; Figure 3).

The diagnoses for both eyes were Marfan syndrome, ectopia lentis, congenital lens coloboma, binocular refractive error (high astigmatism), and anisometropia. She was admitted to the hospital for surgery on her left eye. Phacoemulsification combined with trans-scleral suture fixation intraocular lens (IOL) and anterior vitrectomy was planned. A foldable 3-piece IOL (ZA9003 25.5D, TECNIS, Johnson & Johnson Surgical Vision) was fixed by trans-scleral suture due to the absence of capsular support. The surgical procedure was uneventful. After 3mo, she achieved a vision of 20/30, improving to 20/25 with -0.5 DS correction in the left eye. Her intraocular pressure was 17 mm Hg, and IOL was in the center. The patient was highly satisfied with the improvement in visual quality (Figure 4).

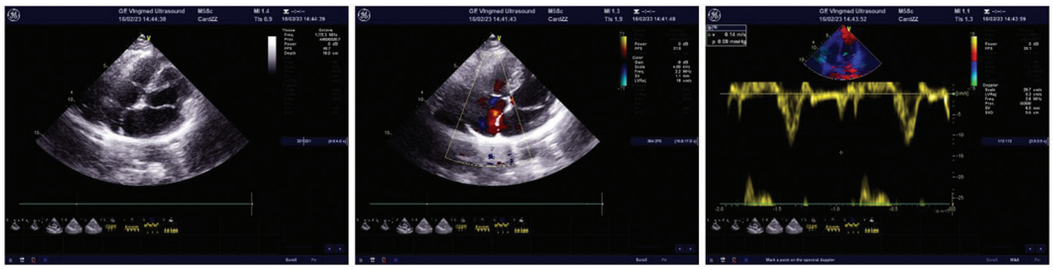

Figure 1 Echocardiography: atrial septal bulging, mild regurgitation of mitral, tricuspid and aortic valves.

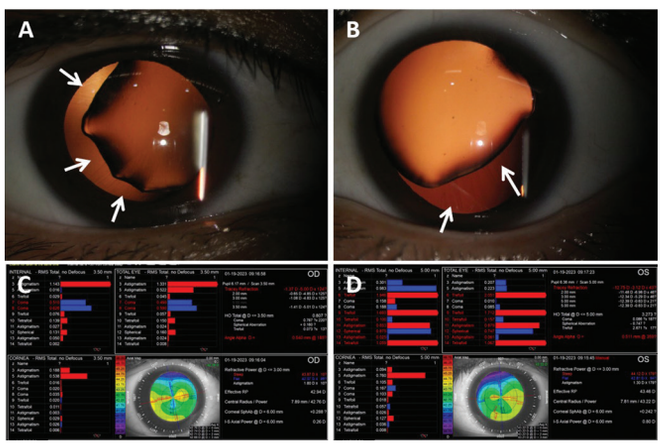

Figure 2 Complaining of decreased vision in both eyes for 3mo A: On slit lamp examination under mydriasis, bilateral ectopia lentis with lens coloboma, along with several notchings and absence of zonules at the equator in the right eye; B: Bilateral ectopia lentis with lens coloboma, along with several notchings and absence of zonules at the equator in the left eye; C: iTrace examination, poor visual function, visual interference mainly from internal eye in the right eye; D: Poor visual function, visual interference mainly from internal eye in the left eye.

DISCUSSION

Lens coloboma is a congenital lens morphological abnormality with an autosomal recessive inheritance. As a kind of ocular coloboma, with an estimated prevalence of 0.7‰. The exact pathogenesis is unkown, however, two theories exist to explain it[3-4]. According to one opinion, the lens is born with aberrant development. Another opinion is that the lens is not a true coloboma, but the faulty development of the zonules (absent or stretched), resulting in the lens retracting to appear notches due to the lack of zonular traction. Both eyes can be involved. The size and form vary due to the differential traction of the zonular fibers. In moderate situations, there is only a tiny notch; however, in severe cases, the defect might reach 1/4 of the lens's equator. The coloboma often occurs at the equator of the lens and can only be found under sufficient mydriasis, which is easy to miss in clinical practice.

Marfan syndrome is a genetic connective tissue disorder affecting many systems, such as the ocular, cardiovascular, and musculoskeletal systems. The incidence rate is 0.1‰-0.2‰. The clinical manifestations and signs are complicated and diversified. Ectopia lentis is the most common ocular symptom, with a 50%-85% incidence rate, which is bilateral and symmetrical. Superior, superotemporal dislocations are most common, while posterior, and superonasal dislocations are relatively less. In our case, the lenses were superonasally dislocated, this maybe due to the congenital inferotemporal lens coloboma. According to previous reports, the dislocation of the lens is progressively aggravated[5]. Additional signs include ametropia, astigmatism, strabismus, amblyopia, glaucoma, flat cornea, hypoplastic iris and ciliary muscle, and others[6].

This case notes: First, the association of lens coloboma with Marfan syndrome has been described in literature, this case indicates that the coexistence of lens coloboma with bilateral ectopia lentis in Marfan syndrome is an uncommon occurrence[4]. Due to the faulty development of the zonules and lens coloboma in the inferotemporal position, the lens subluxated to the opposite side orientation. Second, the round and blunt edge of the lens coloboma area results in an uneven refraction power in different meridian, which can cause excessive astigmatism and ametropia. Third, pupil dysfunction, a poor reaction to the mydriasis medication, and cannot be fully dilated. Histopathology has confirmed that the mechanism is dysplasia or the absence of dilator pupillae muscle. Therefore it is challenging for examination and surgery. Further, the ultrasound biomicroscopy tips that the ciliary body becomes thinner, and the ciliary process is not obvious, which may be related to the dysplasia of the circular muscle of the ciliary body[7]. In the end, due to poor vision, Marfan syndrome is frequently initially identified in eye hospitals. If patients first visit an ophthalmologist, a thorough and systematic examination must be performed, patients must be reminded to pay attention to other relevant symptoms, instructed to screen their children for inherited ocular disorders, and if necessary, referred to other departments (orthopedics, cardiology, and others) for treatment.

Preoperative planning: First, the ultrasound biomicroscopy examination is a valuable test[7]. Second, preoperative mydriasis is very important. Third, inspect the fundus thoroughly to rule out any potential retinal detachment.

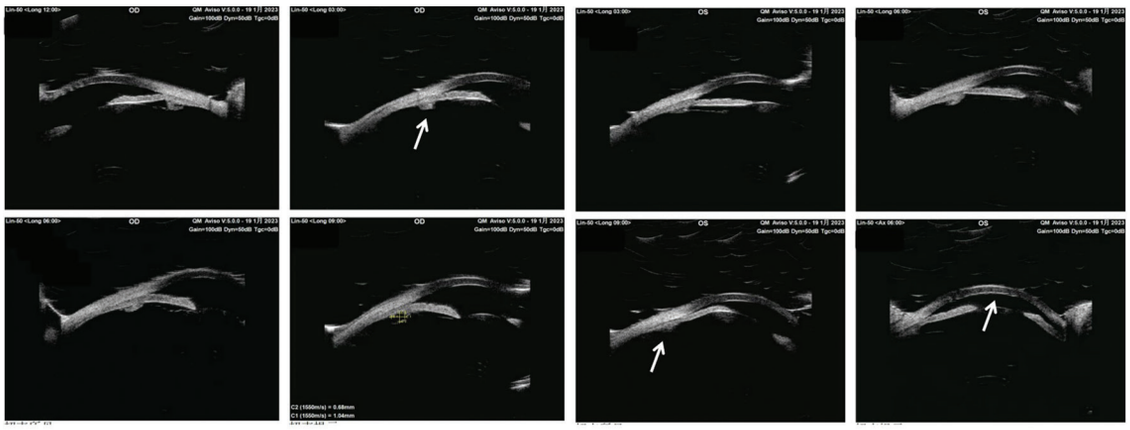

Figure 3 Ultrasound biomicroscopy revealed that anterior chamber depth was uneven, lens subluxation, the ciliary body became thin, and the ciliary process was not obvious.

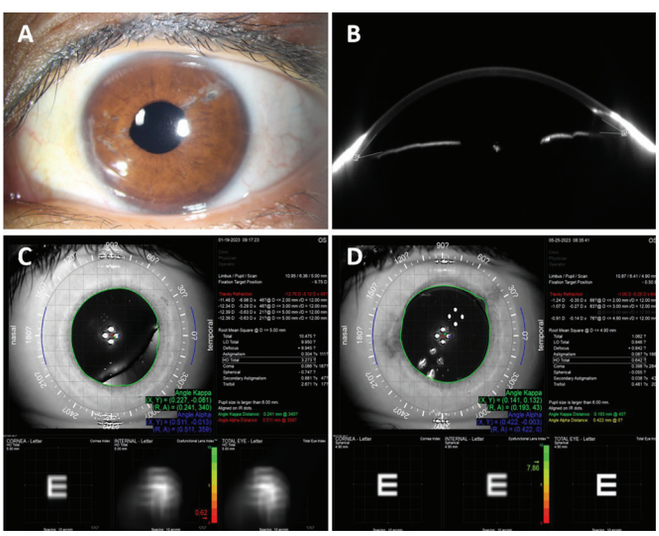

Figure 4 Followed up 3mo A, B: The intraocular lens position was centered; C, D: The comparision before and after surgery showed that the visual quality was improved and the E-sign was clear by iTrace examination.

Fourth, while the len coloboma is large, the risk of vitreous prolapse exists, so anterior vitrectomy equipment should be prepared. Fifth, ophthalmologists have an obligation to remind, especially young patients, of the loss of accommodation after monofocal IOL implantation, except for myopic reservation or presbyopia-correcting IOL implantation. Sixth, emphasize the reconstruction of postoperative visual function.

At present, there are two acceptable interventions. Phacoemulsification combined with scleral fixation 3-piece IOL and capsular tension rings assisted with IOL intracapsular implantation[8]. Considering zonules rupture increased with aging in Marfan syndrome, capsular tension ring is not a good choice in this case. Scleral fixation IOL (with suture or sutureless) is proven to be of good optical outcomes, is relatively stable, and causes less harm to intraocular tissues such as the iris, anterior chamber angle, and corneal endothelium. The IOL is positioned behind the pupil in a physiological position. It's regarded as the best surgical technique for implantation without capsular support[5,9]. Follow-up focuses on items like IOL tilt or displacement due to suture loosening or breakage, pigmentary dispersion, anterior chamber hyphema, intraocular pressure fluctuation, glaucoma, endothelial cell density, vitreous hemorrhage, prolapse of the vitreous, retinal detachment, choroidal detachment, macular edema, endophthalmitis, visual function reconstruction, and others.

In conclusion, the coexistence of lens coloboma with bilateral ectopia lentis in Marfan syndrome is a rare case. Marfan syndrome, as a multi-system disease, requires multi-disciplinary and multi-center cooperation, detection, and treatment. Ophthalmologists should be able to provide a definitive diagnosis for those patients. Evaluating the general and eye conditions comprehensively, confirming the optimal eye surgery plan, to improve the visual function, and quality of life.

ACKNOWLEDGEMENTS

Foundations: Supported by Hunan Province Enterprise Joint Fund Project (No.2023JJ70040); Young and Middle-Aged Science and Technology Innovation Incubation Project of Aier Eye Group (No.AC2214D01); Free Exploration Program of the Scientific Research Foundation of Aier Eye Group in 2021 (No.AF2102D6); Clinical Research Institute Research Fund Project of Aier Eye Group (No.AR2102D1).

Conflicts of Interest: Zhao ZB, None; **Tang KL,** None; **Ding YX,** None; **Yang JK,** None; **Wang HP,** None; **Ma LW,** None.

REFERENCES

1 Sahu, Yadav R, Gupta S, *et al*. Bilateral ectopia lentis with isolated lens coloboma in Marfan syndrome. *GMS Ophthalmol Cases* 2016;6:Doc14.

2 Gupta G, Goyal P, Malhotra C, *et al*. Bilateral lens coloboma associated with Marfan syndrome. *Indian J Ophthalmol* 2018;66(8):1192.

3 Dhiman R, Kaur L, Sharma N, *et al*. Diverse presentations of ectopia lentis and lens coloboma in Marfan's syndrome. *Oman J Ophthalmol*

2023;16(2):310-313.

4 Bhate M, Motwani D, Murthy SI, *et al*. Congenital anomalies of lens shape. *Taiwan J Ophthalmol* 2023;13(4):479-488.

5 Hoffman RS, Snyder ME, Devgan U, *et al*. Management of the subluxated crystalline lens. *J Cataract Refract Surg* 2013;39(12): 1904-1915.

6 Esfandiari H, Ansari S, Mohammad-Rabei H, *et al*. Management strategies of ocular abnormalities in patients with Marfan syndrome: current perspective. *J Ophthalmic Vis Res* 2019;14(1):71-77.

7 Jia WN, Wang QY, Niu LL, *et al*. Morphometric assessment of the ciliary body in patients with Marfan syndrome and ectopia lentis: a quantitative study using ultrasound biomicroscopy. *Am J Ophthalmol* 2023;251:24-31.

8 Thapa BB, Singh R, Ram J, *et al*. Lens coloboma in one eye and ectopia lentis in the other eye of a patient with Marfan syndrome. *BMJ Case Rep* 2014;2014:bcr2014207112.

9 Ma HL, Li SY, Jiang XN. A clinical study of phacoemulsification and foldable intraocular lens implantation with ciliary groove sutured for lens subluxation in Marfan syndrome. *Chin J Pract Ophthalmol* 2014,32(05):620-623.