CASE REPORT

Lens coloboma in one eye and ectopia lentis in the other eye of a patient with Marfan syndrome

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SUMMARY

We present a case of Marfan syndrome with lens coloboma in one eye and ectopia lentis in the other. A 14-year-old girl reported decreased vision in the left eye. Her visual acuity was 6/24 and counting fingers at 1 m in the right and left eyes, respectively. Her intraocular pressure was 15 mm Hg in both eyes. Evaluation of the right eye on slit lamp biomicroscopy under mydriasis revealed an inferiorly visible flattened and concave crystalline lens equator from 4 to 8 o'clock position along with notching and absence of zonules, suggestive of lens coloboma. Left eye examination revealed a superiorly subluxated lens from 3 to 9 o'clock position and posterior subcapsular cataract. The posterior segment evaluation of both eyes was normal. Her father, aunt and grandfather were of tall stature, characteristic of Marfan syndrome. On systemic evaluation, the patient was diagnosed as Marfan syndrome. After surgical correction she achieved vision of 6/6 in both eyes.

BACKGROUND

Lens coloboma is characterised by the lens tissue notching at the equator.¹ It represents an incomplete lens formation due to failure of the fetal fissure to close completely. It can be seen as a peripheral flattening or indentation of the lens. It is usually inferior-nasal and monocular.¹

By far the most common ocular abnormality seen in patients with Marfan syndrome is ectopia lentis, affecting 50–80% of individuals. It is bilateral and symmetrical.² Angra *et al*³ and Mehrotra *et al*⁴ described the association of lens coloboma with Marfan syndrome. Our case was unique, showing lens coloboma in one eye and ectopia lentis in the other of the same patient of Ghent 1 proven Marfan syndrome.⁵

CASE PRESENTATION

A 14-year-old girl presented with the symptom of painless and progressive decreased vision in the left eye for 2 years.

On ocular examination, her unaided distance visual acuity was 6/24 and counting fingers (CF) at 1 m in the right (OD) and left eye (OS), respectively, which improved to 6/12 and CF at 1 m with spectacle correction. Her intraocular pressure (IOP) was 15 mm Hg bilaterally (OU). Undilated anterior segment evaluation on slit lamp biomicroscopy was normal in the right eye but the patient had a superiorly subluxated cataractous lens in the left eye. Examination of the right eye under mydriasis revealed an inferiorly visible flattened and concave lens equator from 4 to 8 o'clock along with the presence

of typical notching (figure 1A arrow) and absence of zonules, suggestive of lens coloboma. Left eye examination revealed a deep and irregular anterior chamber with superiorly subluxated lens from 3 to 9 o'clock along with posterior subcapsular cataractous changes and absence of notching (figure 1C). The posterior segment of OU was normal. There was no history of ocular trauma or surgery. The patient did not have any detected systemic illness. Family history revealed tall stature and incidence of eye surgery at a young age in the patient's father, aunt and grandfather; two characteristics of Marfan syndrome.

On systemic evaluation in the paediatric ward, our patient was diagnosed as Marfan syndrome.

INVESTIGATIONS

The patient had axial length of more than 26 mm and myopia of more than >10 D bilaterally. Systemic examination revealed arachnodactyly, positive wrist and thumb sign, and upper segment to lower segment ratio <1.05. The rest of the systemic work up was normal.

Management and treatment

The patient underwent pars plana lensectomy and sutureless trans-scleral fixation (TSF) of a hydrophobic foldable acrylic intraocular lens (IOL) assisted with fibrin glue in the left eye. She achieved postoperative best corrected visual acuity (BCVA) of 6/18 in this eye.

Six weeks later, she underwent right eye phacoaspiration with posterior chamber in the bag IOL implantation using a Cionni-modified capsular tension ring. Postoperatively she achieved BCVA of 6/6.

She was advised occlusion therapy with spectacles for amblyopia in the right eye. At 1 year follow-up her BCVA is 6/6 bilaterally with well-centred IOL (figure 1B, D).

OUTCOME AND FOLLOW-UP

At 1 year follow-up her BCVA is 6/6 in both eyes, with normal IOP and well-centred IOL.

DISCUSSION

Marfan syndrome is known to present with many ocular abnormalities. Of these, by far the most common, occurring in 50–80% of affected individuals, is ectopia lentis, which is bilateral and symmetrical.² Other manifestations include myopia, retinal detachment, strabismus, glaucoma, flat cornea, megalocornea, and hypoplastic iris and ciliary muscle.² Lens coloboma in association with Marfan syndrome is described in the literature by Angra *et al*³ and Mehrotra *et al*.⁴ To the best of

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Figure 1 (A) Preoperative anterior segment photo of the right eye showing typical notch at lens equator (arrow), absence of zonules, and flattened and concave surface at lens equator. (B) Postoperative photo of the right eye with well-centred intraocular lens in the capsular bag and Cionni ring providing equatorial support with a hook in the sulcus (arrow). (C) Preoperative anterior segment photo of the left eye with superiorly subluxated lens (arrow) with posterior subcapsular cataract. (D) Postoperative photo of the left eye with well-centred intraocular lens.

our knowledge, Marfan syndrome with ectopia lentis in one eye and lens coloboma in the other has never been described.

The management of lens coloboma involving 120° or more is phacoaspiration with IOL using a Cionni-modified capsular tension ring.⁶ Gurler *et al*,⁶ in their prospective study, suggested that phacoaspiration and IOL with capsular tension ring or Cionni ring implantation in isolated lens coloboma was an effective and safe option for providing a refractive correction and significant visual improvement in these cases.

The management of severe subluxated lens, where the lens edge uncovers greater than 50% of the pupil, is difficult. Currently, pars plana lensectomy with anterior vitrectomy and TSFIOL assisted with fibrin glue is the surgical procedure of choice.⁷ Other modalities for management of severe lens subluxation include phacoaspiration and IOL with capsular tension ring or Cionni ring, Ahmed capsular tension segment implantation,⁸ pars plana lensectomy with vitrectomy and sutured TSFIOL.⁹

Learning points

- ▶ Lens coloboma and subluxation can inadvertently be missed, so examination after mydriasis is essential.
- ▶ Lens coloboma can mistakenly be diagnosed as lens subluxation; typical notching, concavity of the lens equator and absence of zonules are diagnostic feature of lens coloboma.
- ▶ Lens coloboma associated with Marfan syndrome has been shown in earlier reports; our report adds more evidence by proving this association with Ghent 1 nosology.

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Contributors BBT wrote the manuscript, collected the references, revised the manuscript and approved the finalised manuscript. RS was involved in the planning and writing of the manuscript, and approved the finalised manuscript. JR provided the concept, revised the manuscript and approved the finalised manuscript. AK was involved in writing and revision of the manuscript. All authors were involved directly in the management and follow-up of the patient.

Competing interests None.

Patient consent Obtained.

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