Case report

A new combined surgical approach in a patient with microspherophakia and developmental iridocorneal angle anomaly

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Abstract

Backround: We describe a patient with microspherophakia due to Weill-Marchesani syndrome accompanied by developmental angle anomaly who was successfully treated with a surgery of combined PPL and goniotomy. **Case:** We report ocular findings of a 1-year-old girl who was diagnosed with Weill-Marchesani syndrome with a positive family history of glaucoma in her cousins and glaucoma with anterior segment dysgenesis in her older brother. Anterior segment examination revealed clear corneas with 13 mm horizontal diameter in the right and 12.5 mm in the left and very shallow anterior chambers centrally and peripherally in both eyes. Although axial lengths were 18.9 mm in the right and 19.1 mm in the left eye, cycloplegic refractive errors were -7.75 DS (-2.75 at 1800) in the right eye and -8.50 DS (-2,75 at 1800) in the left eye. Intraocular pressures were 34 mmHg in the right and 38 mmHg in the left eye. **Observation:** Following pars plana lensectomy, gonioscopy revealed developmental iridocorneal angle anomaly and goniotomy was performed at the same session. During 3 years of follow-up, the patient experienced no complications. IOP was 12 mmHg without medication in the last visit; cup-to-disc ratio and corneal diameters were stable. **Conclusion:** The coexistent microspherophakia and developmental iridocorneal angle anomaly can be successfully treated with combined pars plana lensectomy and goniotomy.

Keywords: Microspherophakia, developmental iridocorneal angle anomaly, pars plana vitrectomy and lensectomy, goniotomy

Introduction

The Weill-Marchesani syndrome is an infrequent alteration of the connective tissue which involves the eyes. Patients exhibit low height, brachydactyly with joint stiffness, progressive myopia, microspherophakia, lens luxation and secondary glaucoma (Veiga de la Jara C et al, 2006). In microspherophakia, the lens is small and spherical. An abnormality in the zonular fibers would allow the lens to remain spherical instead of tending to a bi-convex shape (Veiga de la Jara et al, 2006). The tendency towards dislocation and displacement of the lens, would explain the progression of lenticular myopia and narrowing of the anterior chamber and closure of the angle (Veiga de la Jara et al, 2006 and Willoughby and Wishart, 2002).

Different surgical management techniques comprises YAG-laser peripheral iridotomy (Bhattacharjee et al, 2010), lens extraction

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with phacoemulsification with or without intraocular lens (IOL) and capsular tension ring implantation and anterior vitrectomy (Willoughby and Wishart, 2002; Bhattacharjee et al, 2010), pars plana lensectomy (PPL) and anterior vitrectomy (Kaushik S et al, 2006), goniosynechiolysis and lensectomy (Kanamori A, 2004) and trabeculectomy (Asaoka et al, 2003 and Yasar, 2003) for glaucoma in microspherophakia.

Here we describe a patient with microspherophakia accompanied by developmental angle anomaly who was successfully treated with a surgery of combined PPL and goniotomy.

Case

A one-year-old girl was brought to our clinic by her parents, with complaints of big eyes that had been noticed since the child's birth and they also wanted to check her eyes because of their son's eye disease.

The child was born full term by normal vaginal delivery with no history of acute illness or drug use by the mother. The child's parents were second cousins. Her family history was notable for glaucoma which had been diagnosed in her two cousins.

The patient's older brother was 4-year-old. He has short stature (below third percentile) and brachydactyly. His retinoscopic examination revealed -30 Diopter myopia in both eyes. Slit-lamp examination of his eyes showed anterior segment dysgenesis findings, including posterior embryotoxon, peripheral iridocorneal adhesions, iris pathology ranged from mild stromal thinning to marked atrophy in various segments in both eyes (Figure 1a, 1b). The anterior chamber was shallow in the left eye. Central corneal opacity and anterior synechia was seen in his right eye (Figure 1a). Dilated slit-lamp examination revealed bilateral microspherophakia showing the increased anteroposterior diameter of the lens. Anterior segment optical coherence tomography confirmed that the lens had decreased equatorial diameter with increased antero-posterior diameter (Figure 2). Corneal diameters were 13 mm in the right eye and 12,5 mm in the left eye, horizontally. Intraocular pressures (IOP) were 16 mmHg in the right eye and 12 mmHg in the left eye with antiglaucoma therapy.

Our patient's physical examination was notable for short stature (below third percentile) and brachydactyly, too.

On external examination of eyes, the patient appeared to have megalocorneas and very shallow anterior chamber. The lens of both eyes appeared to be microspherophakic on B-mode ultrasonography (Figure 3).

An anterior segment examination with a surgical microscope revealed bilateral clear cornea and very shallow anterior chamber centrally and peripherally.

Cycloplegic refractive error was -7.75 DS (-2.75 at 180^0) in the right eye and -8.50 DS (-2.75 at 180^0) in the left eye. Keratometric values were 7.55/8.14 mm in the right eye, 7.47/8.17 mm in the left. Axial length measured with A-mode ultrasonography was 18.9 mm in the right and 19.1 mm in the left eye. Horizontal corneal diameters were 12.5 and 13 mm in the right and left eye, respectively. Central corneal thickness with ultrasonographic pachimetry was 530 μ in the right eye, 536 μ in the left eye. IOP obtained via Tono-Pen XL applanation tonometer (Reichert Inc. NY, USA) was 34 mmHg in the right and 38 mmHg in the left eye. Fundus examination showed no abnormal findings.

In the light of these findings, a tentative diagnosis of Weill-Marchesani syndrome was made. Chromosomal studies could not be performed on either the patient or her brother because of the lack of appropriate facilities at our center and financial issues of family.

Bilateral surgical removal of the lens and intraoperative gonioscopic examination were

planned. Gonioscopic examination showed the high iris insertion and trabeculodysgenesis in both eyes. The patient underwent combined PPL, vitrectomy and nasal 160^0 goniotomy in both eyes with a 1-week interval.

Routine topical antibiotic and steroid medication and pilocarpine were given post-operatively for 2 months. Following surgery the IOP was decreased to low 10s without medication with deep anterior chamber and clear corneas. The patient was given +15.0 diopter glasses.

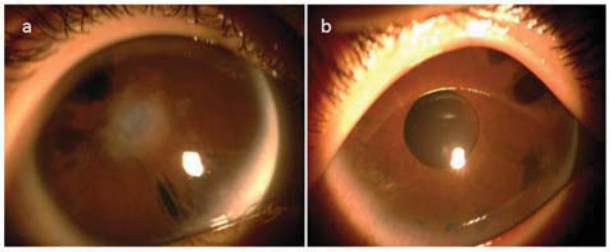

Figure 1: The biomicroscopic appearance of the eyes of the patient's brother (1a: Right eye, 1b: Left eye)

Figure 2: Anterior segment optical coherence tomography appearance of the lens with decreased equatorial diameter and increased antero-posterior diameter

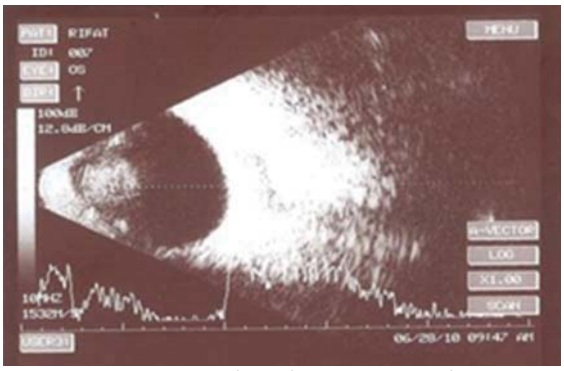

Figure 3: B-mode ultrasonography revealed microspherophakic lens and shallow anterior chamber in both eyes.

Figure 4: Anterior segment appearance of patient at 3 years control.

During 3 years of follow-up the patient experienced no complications. IOP was 12 mmHg without medication in the last visit; cup-to-disc ratio and corneal diameters were stable in both eyes (Figure 4).

Discussion

Anterior segment dysgenesis is caused by failure of neural crest cell migration and differentiation (Rastogi et al, 2010). Thut et al reported that mutations in lens tissue can result in abnormalities in iris and ciliary body of mice (Thut et al, 2001). The lens, being ectodermal in origin, the association of microspherophakia cannot be explained with anterior segment dysgenesis of neural crest origin. Abnormal lens development is probably the primary defect that failed to provide the signal to the surrounding developing anterior segment structures (Thut et al, 2001). To our knowledge, only one patient having both microspherophakia and Axenfeld-Rieger syndrome has been previously described in the literature (Rastogi et al, 2010).

IOP elevation in spherophakia can occur by a variety of mechanisms (Willoughby and Wishart, 2002; Kaushik et al, 2006; Asaoka et al, 2003). Angle-closure glaucoma occurs in spherophakia from a pupillary block mechanism caused by dislocation or forward movement of the lens, depending on the zonular integrity. When the zonules are intact, the anterior lens

curvature and forward positioning of the lens predispose to iridolenticular contact and pupil block. Unrelieved pupillary block and iris bowing can lead to peripheral anterior synechia (PAS) formation and irreversible trabecular damage (Willoughby and Wishart, 2002). Acquired or congenital abnormalities of the trabeculum have also been described in eyes with Weill-Marchesani syndrome (Feiler-Ofry et al, 1968). In our patient, both the pupillary block mechanism related to microspherophakia and the high iris insertion and iridocorneal angle abnormality could be valid for mechanisms of glaucoma.

The management of glaucoma in spherophakia is still controversial. Willoughby CE and Wishart PK (2002) described a case of spherophakia with glaucoma whose IOP could be controlled following lensectomy. However, Yasar (2003) described a patient in whom lensectomy could control the IOP in the short-term, but who subsequently required trabeculectomy. Asaoka et al (2003) reported successful control of IOP with trabeculectomy in a patient with spherophakia and open angles . Kaushik et al (2006) reported that pupillary block can be relieved by lensectomy, but it may not suffice to control the IOP in the presence of extensive PAS. Kanamori et al (2004) reported good IOP control with goniosynechialysis with lens aspiration and IOL implantation in a patient with spherophakia and chronic angle closure glaucoma .

For our patient's older brother, the anterior segment dysgenesis findings gave rise to think of Axenfeld-Rieger anomaly in the left eye and Peters anomaly in the right eye. Based on this finding and the presence of increased corneal diameters in our patient, we needed to examine the iridocorneal angle after lens extraction. Due to the presence of trabeculodysgenesis we decided to combine the goniotomy procedure with PPL. To our knowledge, the use of this procedure in patient with microspherophakia and developmental iridocorneal angle anomaly has not been previously described in the literature.

Conclusion

Bilateral combined goniotomy and PPL procedure can be used in patients with microspherophakia and secondary glaucoma due to developmental angle anomalies.

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