deprivation amblyopia. With bilateral idiopathic congenital cataracts, the posterior segment is often spared, and early cataract removal is recommended for the best visual outcome with timeline to initial surgery between 4-6 weeks for unilateral cataracts and 4-10 weeks for bilateral cases.[9] A 50% glaucoma risk reduction has been reported if cataract surgery is delayed from 4 to 8 weeks of age as opposed to a threefold higher risk in those eyes undergoing early removal between 4-6 weeks of age (compared to delay in surgery from 7 weeks to 6 months).[10] In light of reported cases of childhood glaucoma in CDG patients, if limited visual potential secondary to irreversible causes, such as retinal or optic nerve pathology, were known prior to cataract surgery, both parents and physician may have weighed the risks/benefits of early versus late surgery differently given the risk of lifelong glaucoma.[9]

The risks of anesthesia, especially in syndromic patients with multisystem dysfunction, should also be weighed carefully. We were able to combine cataract surgery with other medically necessary procedures to limit the overall anesthetic burden to our patient.

Our experience may help parents and physicians to make informed decisions regarding treatment of CDG and timing of cataract surgery, given the potential for retina and/or optic nerve abnormalities that may limit the final visual outcome. Unilateral cataract surgery to limit both anesthetic and bilateral glaucoma risks, once such posterior segment pathology is discovered, may also be considered in these cases.

Literature Search

PubMed MEDLINE, OVID MEDLINE, and Clinical Key were searched in December 2018, without date restriction, using the following search terms: DPAGT1, *congenital glycosylation disorder*, *CDG*, *congenital disorders of glycosylation*, *congenital myasthenia syndrome*, *CMS*, *ocular findings in CDG*, *ocular findings in CMS*, *retina* AND *CDG* or *retina* OR *CMS*, *eye* AND *CDG* OR *CMS*, *congenital cataract* AND *glaucoma* OR *amblyopia*. Articles cited in reference lists of retrieved articles were also researched. Foreign-language literature was considered when applicable.

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Pigment dispersion syndrome and response to laser peripheral iridotomies in a child with Marfan syndrome

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Pigment dispersion syndrome (PDS) and pigmentary glaucoma have rarely been reported in Marfan syndrome and have never been reported in a child with Marfan syndrome. We report the clinical and ultrasound biomicroscopic findings of PDS in a 14-year-old girl with Marfan syndrome and its favorable response to bilateral laser peripheral iridotomy.

Marfan syndrome is a systemic connective tissue disorder that results from mutations in the *FBN1* gene that encodes fibrillin-1.[1] Ocular

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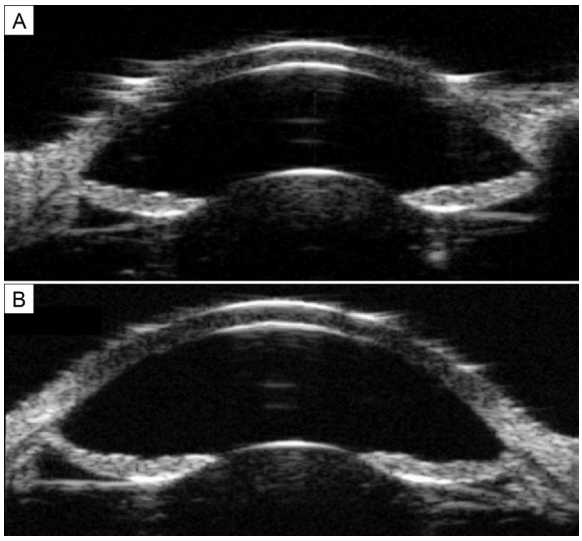

FIG 1. Ultrasound biomicroscopy (UBM) of the anterior chamber of each eye at presentation showing a back-bowed iris configuration with iridozonular apposition.

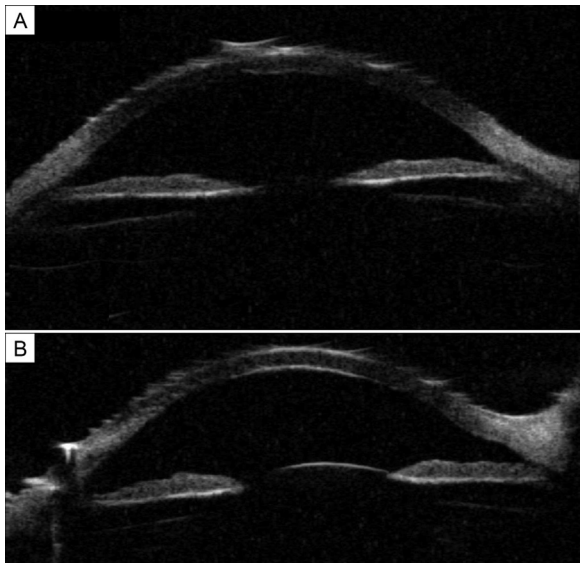

FIG 2. UBM of the anterior chamber of each eye after laser peripheral iridotomy showing restoration of normal, flat iris configuration and resolution of iridozonular apposition.

manifestations of Marfan syndrome include ectopia lentis, high myopias, rhegmatogenous retinal detachments, open-angle glaucoma, and angle-closure glaucoma secondary to lens subluxation. Of note, fibrillin has been documented diffusely within iris stromal tissue, particularly anterior to the pigmented layers close to the dilator and surrounding the sphincter muscles.[2]

Pigment dispersion syndrome (PDS) is a disorder of iridozonular apposition that can lead to pigmentary glaucoma (PG), a secondary open-angle glaucoma caused by progressive trabecular dysfunction related to pigmentary release. In the US, the annual incidence of diagnosed PDS is 5/100,000; of PG, 1.5/100,000. Pigment release accompanied by IOP elevation has been observed after strenuous exercise in some patients. Risk factors include male sex (PGC 2:1-5:1, PDS 1:1), age (most common in men in their 30s and women in their 40s, although PDS in a 12-year-old has been reported),[3] myopia (usually moderate), white race, concave iris and posterior iris insertion, flat corneas, and positive family history. Laser peripheral iridotomy (LPI) is sometimes performed in these cases in an attempt to limit pigment dispersion. A Cochrane systematic review in 2016 concluded that there is "no clear benefit for LPI compared to no laser in eyes with PDS or pigmentary glaucoma (PG) in preventing VF loss."[4] The authors of the review did, however, note few adverse effects of LPI and "very low quality of evidence" for LPI. There were no incidents of retinal detachment. A recent metaanalysis published by the American Academy of Ophthalmology reported complications of LPI in patients with primary angle-closure glaucoma: transient IOP spike (6%-10% with IOP elevation of at least 8 mm Hg from baseline), dysphotopsia (2%-11%, less with temporal compared with superior LPI), anterior chamber bleeding (30%-41%), cataract progression (23%-39% over a follow-up period of 1-6 years), need for repeat LPI (1-2%), and transient anterior chamber inflammation.[5] There were no cases of retinal detachment. Scott and colleagues[6] performed the only prospective study of LPI for the prevention of pigment dispersion glaucoma in the literature and reported no incidents of retinal detachment over 3 years. Although Scott's group observed no benefit from LPI, patients in this study already had both pigment dispersion and ocular hypertension, and the authors noted that "treatment may be effective in younger patients: those without irreversible trabecular meshwork damage or in those with documented increased iridozonular contact (iris concavity and more posterior iris root insertion)." We describe the clinical and echographic findings of PDS before and after LPI in a young patient with Marfan syndrome.

Case Report

A 14-year-old white girl presented at the Minnesota Lions Children's Eye Clinic at the University of Minnesota with a chief complaint of slowly progressive blurry vision. She had been through multiple rounds of glasses and toric contact lenses, with unsatisfactory correction. Her past medical history was significant for genetically confirmed Marfan syndrome (c.3476G>A, p.Cys1159Tyr) with progressive aortic root dilation and mitral valve prolapse. She was being treated with low-impact activity restriction, losartan, and atenolol. On examination, best-corrected visual acuity was 20/40 −2 in the right eye, with refraction −1.50 +1.50 ×040, and 20/50-2 in the left eye, with refraction −1.25 +1.00 ×070. Retinoscopy revealed additional irregular astigmatism that was unable to be corrected. Intraocular pressure (IOP) was

18 mm Hg in each eye by Icare tonometry (Icare USA, Raleigh, NC). Her pupils were equally reactive, with no afferent defect, and her visual fields were full. She was orthophoric at distance, with an exophoria of 4$^{\Delta}$ at near, and had full ocular motility. Slit-lamp examination of both eyes was remarkable for 3+ endothelial pigment centrally in a "Krukenberg spindle" conformation with 3+ free pigment floating in a deep anterior chamber prior to dilation. It would have been classified as 4+ pigment, given the extreme density of free-floating pigment, but no pile of pigment was evident in the angle. Gonioscopy of both eyes using Spaeth grading revealed D45q with 4+ PTM (pigmented trabecular meshwork) and a Sampaolesi's line for 360° in each eye. There was diffuse fine pigment stippling on the anterior iris surface and marked peripheral transillumination defects in both eyes (eSupplement 1A, available at jaapos.org). Dilated examination was remarkable for mild lens subluxation, with pigment staining on the zonules and a dense Scheie stripe on the equatorial lens capsule in both eyes (eSupplement 1B). In each eye, the vitreous was clear, the optic disk was pink and sharp (0.1 cup:disk ratio), and the macula and periphery was normal. Static perimetry using the Octopus visual field G-TOP protocol (Haag-Streit USA, Mason, OH) was normal. Anterior segment ultrasound biomicroscopy (UBM) was notable for a back-bowed iris configuration with iridolenticular and iridozonular apposition in each eye (Figure 1). The patient was diagnosed with pigment dispersion syndrome with normal IOP and without evidence of glaucomatous optic neuropathy.

Given the florid amount of active pigment dispersion and UBM findings, laser peripheral iridotomy was recommended and performed first on the right eye with near-total reduction of pigment dispersion, and then on the left eye (eSupplement 1C). Postoperative examinations were remarkable for stable visual acuity, normal IOP, near-resolution of circulating anterior chamber pigment (graded as "trace" by the examiner), and restoration of normal iris conformation on repeat UBM in both eyes (Figure 2). One year later, the patient's examination was stable, with absence of any free-floating pigment on anterior chamber examinations in both eyes.

Discussion

Three cases of pigment dispersion in Marfan syndrome have been reported in the literature. The first was a 34-year-old man with asymmetrical pigmentary glaucoma secondary to significant asymmetrical crystalline lens subluxation, which was controlled on timolol and latanoprost.[7] The second was a 37-year-old white man with bilateral pigmentary glaucoma; treatment was not discussed.[8] The third was a 22-year-old Asian-Indian man with bilateral advanced glaucoma with findings of pigment dispersion syndrome who presented with peripheral iridotomies in both eyes.[9]

Our case is unique in its adolescent age-of-onset, florid pigment dispersion bilaterally, ultrasound biomicroscopic imaging of the iris configuration, and good response bilaterally to LPI. We recommend screening adolescent patients with Marfan syndrome for PDS with careful slit-lamp examination. LPI may be considered when there is evidence of both significant pigment dispersion and iris back-bowing to reduce the burden of pigment dispersion in hopes of preventing the development of pigmentary glaucoma.

The risk of retinal detachment in patients with Marfan syndrome is between 4% and 26%.[10,11] There are no reports of retinal detachment after LPI in the literature in other patient populations; however, given the already high risk of retinal detachment in patients with Marfan syndrome, the informed consent conversation should clearly articulate and balance the theoretically increased risk of retinal detachment after LPI and the possible prevention of pigmentary glaucoma. Preoperative screening and postoperative surveillance should include a dilated fundus examination with scleral depression.

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