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Case Report

Juvenile Open Angle Glaucoma: A Report of Three Sons in One Family

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Abstract

A male child aged 16 years old diagnosed with bilateral juvenile open angle glaucoma (JOAG) underwent trabeculectomy with mitomycin C (MMC) in both eyes. For 5 years follow-up, no complications were recorded, and the intraocular pressure (IOP) was well controlled. Family screening was done and revealed JOAG in his brother, 13 years old, and his sister, 9 years old. Both underwent trabeculectomy with MMC. Ologen implant was intraoperatively added in the younger sister. Examination of the parents revealed that their mother, 45 years old, had mild primary open angle glaucoma and medical treatment was prescribed for her with good IOP control, stable perimetry and retinal nerve fiber layer thickness for 4 years follow-up.

Keywords: Juvenile Open Angle Glaucoma; Mitomycin C; Ologen; Trabeculectomy

Introduction

Juvenile open angle glaucoma (JOAG) differs from primary open angle glaucoma (POAG) in the age of onset as it includes age group from 3 years to 35 - 40 years and often has extremely high and large fluctuating intraocular pressure (IOP) [1,2]. It is differentiated from late recognized congenital glaucoma by the absence of large globe, Descemet's membrane breaks (Haab's striae), and anterior segment dysgenesis [3]. Most cases are inherited as an autosomal dominant pattern with more than one third of cases are associated with Myocilin (MYOC) gene mutations [4,5]. In a population-based study, the frequency of JOAG was 0.38 per 100,000 inhabitants between the age of 4 to 20 years old [6]. In this report, we describe three brothers discovered to have JOAG, methods of treatment and their follow-up results.

Case Presentation

A male child aged 16 years old presented to the first author for glasses prescription. There was no history of ocular trauma or surgery with no systemic disease or medication. Family history was irrelevant. Best corrected visual acuity was 20/20 in both eyes, refraction was -0.75 sphere in the right eye and -1.25 -0.5 x 135 in the left eye. The anterior segment examination was normal. Funds slit lamp biomicroscopy revealed optic disc cupping in both eyes with superior notch. Gonioscopic examination showed normal open anterior chamber angle with 1+ trabecular pigmentation and straight iris configuration. IOP was 50 mmHg and 44 mmHg in the right and left eyes, respectively. Central corneal thickness in both eyes were 534 µm. Perimetry, optic disc photography and retinal nerve fiber layer (RNFL) are shown in figure 1. Diagnosis of JOAG was confirmed and medical treatment was started, but adequate IOP was not achieved. Trabeculectomy with mitomycin C (MMC) eventually performed in both eyes. For 5 years of follow-up, no complications were recorded and the IOP was well controlled with addition of topical B-blocker twice daily in the right eye 6 months after surgery to reach the target IOP. In the most recent visit, IOP was 11 mmHg

in the right eye and 12 mmHg in the left eye with stable perimetry and RNFL thickness. Family screening was done and revealed JOAG in his brother, 13 years old and his sister, 9 years old. Both underwent trabeculectomy with MMC. Ologen implant was intraoperatively added in the younger sister. Examination of the parents revealed that their mother, 45 years old, had mild POAG and medical treatment was prescribed for her with good IOP control, stable perimetry and RNFL thickness for 4 years follow-up.

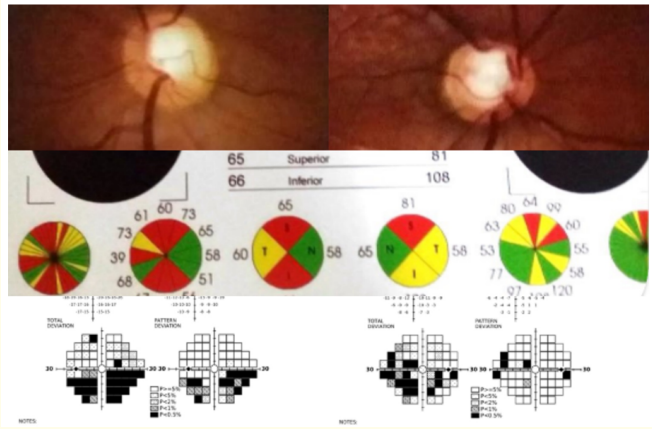

Figure 1: Fundus photos show disc cupping and superior notch in the right and left eyes (upper panel). OCT images show evident retinal nerve fiber layer thinning in the superior and inferior quadrants of the right eye and the superior quadrant of the left eye (middle panel). Visual field shows lower arcuate defect in the left eye and paracentral scotoma in the right eye (lower panel).

Discussion

JOAG is a rare form of glaucoma that differs from adult-onset POAG in its age of onset and often in the magnitude of IOP elevation [1,2]. The genetic basis of JOAG is much more obvious than that of POAG. JOAG is frequently passed down through families as an autosomal trait [4,5]. Many cases of JOAG are caused by mutations in the myocilin gene, especially subjects that have early-onset of disease, high IOP, and a strong family history of glaucoma. As many as 8 - 63% of JOAG cases are associated with MYOC mutations [7]. In this case report, although genetic study could not be done, the early onset and the familial presentation strongly suggest the presence of genetic mutation.

Although adult-onset POAG usually does not follow simple Mendelian genetics and is etiologically complex, genome-wide association studies have shown genetic susceptibility. In some cases, it can be caused by heterozygous mutation in MYOC, OPTN, or WDR36 [4]. In addition, in 2009, heterozygous NTF4 mutation was associated with the phenotype in a small percentage of patients from a German cohort [8]. So screening of parents of JOAG children as well as children of POAG patients may be helpful for early detection of both conditions.

Once the patient has been diagnosed with JOAG, management involves lowering IOP and assessing the rate of progression with serial optic nerve head imaging and static automated perimetry. In JOAG cases, the IOP is often refractory to maximal tolerated medical therapy and incisional surgery is needed. Trabeculectomy with MMC remains the gold standard glaucoma surgery. In this case report, it was used for the 3 brothers, and success was maintained through 5 years of follow-up and without serious postoperative complications. However, given the younger age range of JOAG patients and their life expectancy, it is worth considering an alternative to trabeculectomy with antifibrotics, which carries a risk of up to 1.2% per year of bleb-associated endophthalmitis [9]. The Trabectome or a trabecular bypass shunt may be considered as a conjunctiva-sparing surgical option that is targeted to the disease, especially since JOAG patients may have a genetically maldeveloped trabecular meshwork. Outcomes with the Trabectome have been promising in a small case series [10].

Conclusion

This case report describes diagnosis and treatment of 3 brothers with JOAG. Additionally, their mother was diagnosed with POAG. Such findings confirm the genetic basis of JOAG and suggest a possible relation between POAG and JOAG.

Key Messages

Early screening of other members of the family (irrespective of age) with a known glaucoma patient can help to diagnose JOAG at an earlier stage and could probably prevent or postpone the final visual deterioration.

Source(s) of Support

None.

Conflicting Interest

None.

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