

Clinical Characteristics of Juvenile-Onset Open Angle Glaucoma

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ABSTRACT

Juvenile open-angle glaucoma (JOAG) is an uncommon form of primary open-angle glaucoma, with earlier onset (3 to 40 years of age), higher IOP⁽¹⁾, and more severe visual field loss compared with adult onset primary open-angle glaucoma (POAG). Many studies report that this form of glaucoma typically demonstrates an autosomal dominant inheritance pattern. The myocilin (MYOC) gene is identified abundantly through linkage analysis in the trabecular meshwork (TM) of the affected patients.^{(2) (3)} In this study, we retrospectively demonstrated the clinical characteristics in patients with JOAG, to provide medical treatment and future prevention and awareness in clinics of this disease. Patients with JOAG are diagnosed at an early age and therefore have a longer expectancy than the typical glaucoma patient. And this can cause visual impairment and blindness which can significantly impair the patient's quality of life and limit daily living activities. This retrospective study at Dr. Shroff's Charity Hospital from April 2019 to December 2020 included 80 glaucoma patients, revealing 3.4% with juvenile-onset open-angle glaucoma (JOAG). Notably, 92.5% had bilateral JOAG, and 75% were male. Most patients (87%) lacked a family history of glaucoma, and 91.10% had no systemic diseases. Chief complaints included blurring of vision (38%), ocular pain (9%), and various others. Refractive analysis showed 46% mild myopia, 23% moderate myopia. Visual impairment was absent in 83% of eyes, and cupping severity varied. Disc size was mostly medium (75%), and rim loss severity varied. Among 154 eyes, 131 received medical and surgical treatment, with a mean intraocular pressure (IOP) of 19.2105 mm Hg. The remaining 23 patients had a mean IOP of 19.133 mm Hg. These findings provide concise insights into JOAG patient characteristics and treatment patterns.

Keywords: Juvenile open-angle glaucoma (JOAG), Juvenile onset open-angle glaucoma, Glaucoma

INTRODUCTION

Glaucoma is a neurodegenerative disorder characterized by the progressive optic neuropathy and visual field loss, for which intraocular pressure (IOP) is the chief modifiable risk factor. Glaucoma is the most common cause of irreversible blindness.

Juvenile open-angle glaucoma (JOAG) is an uncommon form of primary open-angle

glaucoma, with earlier onset (3 to 40 years of age), higher IOP⁽¹⁾, and more severe visual field loss compared with adult onset primary open-angle glaucoma (POAG).

The overall prevalence of POAG ranges from 1.0% to 3.9%, depending on the race, age and definitions used [1-3]. Juvenile-onset open angle glaucoma (JOAG) is an uncommon subset of POAG characterized by an

autosomal dominant pattern of inheritance [4,5]. It generally affects individuals during childhood or early adulthood but is distinct from congenital glaucoma that presents with buphthalmos, megalocornea, Haab's striae, and ocular or other systemic developmental anomalies.

Many studies report that this form of glaucoma typically demonstrates an autosomal dominant inheritance pattern. The myocilin (MYOC) gene is identified abundantly through linkage analysis in the trabecular meshwork (TM) of the affected patients.⁽²⁾⁽³⁾

An important ocular finding that had been associated with JOAG is myopia.⁽⁴⁾⁽⁵⁾⁽⁶⁾ JOAG is differentiated from late congenital glaucoma and other childhood glaucoma by the absence of Buphthalmos, Haab's striae and anterior segment dysgenesis.⁽⁶⁾ A recent population-based study reported that the incidence of JOAG was 0.38 per 100,000 residents between 4 to 20 years of age.⁽⁶⁾ based on a study from the Dallas Glaucoma registry, which comprised 376 eyes of 239 childhood glaucoma patients, 4 % of patients has Juvenile open- angle glaucoma.⁽⁷⁾

Juvenile open- angle glaucoma often occurs as a result of abnormal development of trabecular meshwork.⁽⁸⁾ Therefore, drainage of aqueous humor may not be adequate, leading to increased intraocular pressure and damage to the optic nerve.⁽⁹⁾

In this study, we retrospectively demonstrated the clinical characteristics in patients with JOAG, to provide medical treatment and future prevention and awareness in clinics of this disease. Patients with JOAG are diagnosed at an early age and therefore have a longer expectancy than the typical glaucoma patient. And this can cause visual impairment and blindness which can significantly impair the patient's quality of life and limit daily living activities.

Risk Factors

Juvenile-onset open-angle glaucoma has many risk factors such as:

- Male sex: in a study of 125 eyes with JOAG, 64 % were male.⁽⁵⁾

- Myopia.
- Severe elevation of IOP: it is a typical feature of juvenile-onset primary open – angle glaucoma.
- African ancestry.⁽¹⁰⁾

MATERIALS AND METHODS

This retrospective study was carried out at Dr Shroff's Charity Eye Hospital, Daryaganj (A Tertiary Eye Hospital). Before being enrolled in this study, every patient had a thorough ophthalmic examination. 154 eyes belonging to 79 patients with JOAG were examined from April 2019 to December 2020. The only eyes that were taken into account were those with JOAG. Data collected from the medical records of patients diagnosed with JOAG was the basic demographic data included the Age, Gender, Laterality, complaints, family history, refractive error, clinical characteristics of the patients which included the presenting best corrected visual acuity, IOP, the Cup Disc ratio, Disc characteristics, clinical features and OCT, Visual field report interpretation were recorded. The evaluated medical history included patient's family history of glaucoma, gender, IOP, chief complaint at the initial visit Initial evaluation of patients include Visual Acuity was measured by using Snellen's chart. C: D ratio was taken by using 90D lens and Intra Ocular Pressure (IOP) was measured using Goldmann Applanation Tonometer only. Gonioscopy was performed by 4 mirror gonioscope with Slit Lamp Bio microscope. The diagnosis of JOAG was based on the following-

- The presence of characteristic glaucomatous optic nerve head change.
- Visual field defect not attributable to other causes.
- Open anterior chamber angle in the affected eye on Gonioscopy.

All patients with unilateral or bilateral juvenile open angle glaucoma were included in this study who visited in the time period between April 2019 to December 2020.

Excluded were

- Patients with features suggestive of secondary causes of glaucoma based on history or findings from examination.
- Patients those who were having drop out in the follow up visits
- Patients whose investigations were not done, was also excluded from the study

RESULT

As this was a hospital based retrospective study, therefore it was seen that the total

number of Glaucoma patients presented at Dr Shroff's Charity Hospital from the time period between April 2019 to December 2020 was 4763, out of which patients with JOAG were about 161 (i.e, 3.4%). In this study we enrolled 80 Patients At initial hospital visit, 6 patients (7.5%) were diagnosed with unilateral JOAG and 74 patients (92.5%) were diagnosed with bilateral JOAG. (fig 1) 75% of patients were male and 25 % were females. (fig. 2)

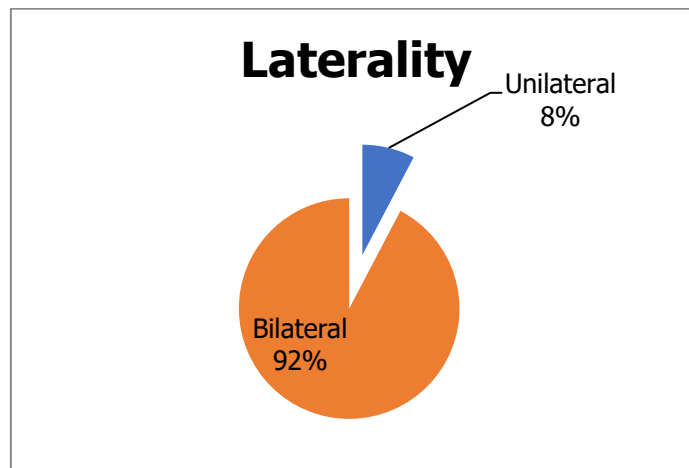


Fig:1

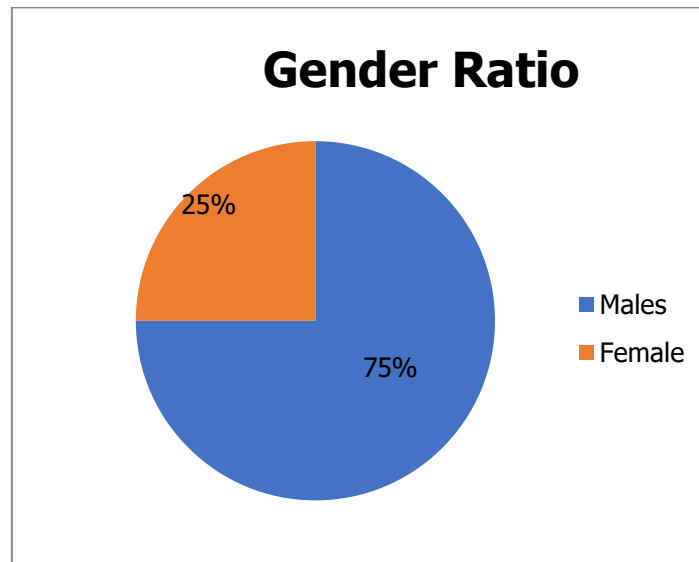


Fig:2

It was found that 87% of the patients were either unaware about their family history or did not have any and rest 13% had a positive family history, it was also found that 91.10 % Patients did not have the history of systemic

diseases like diabetes, thyroid, hypertension, cardiac, asthma etc.

38% of patients came with the chief complaint of blurring of vision, 31% of the patients did not have any complaints, 9% had ocular pain, 5% had heaviness, 5% came

with headache, 4% had redness, 4% had itching, 4% had watering. (fig:5) When analysed for refractive status we found that 46% of eyes were having mild myopia, 23%

had moderate myopia, 21% had high myopia, 6% had mild hyperopia, whereas the percentage of emmetrope and high hyperope was 2 % each. (fig 6)

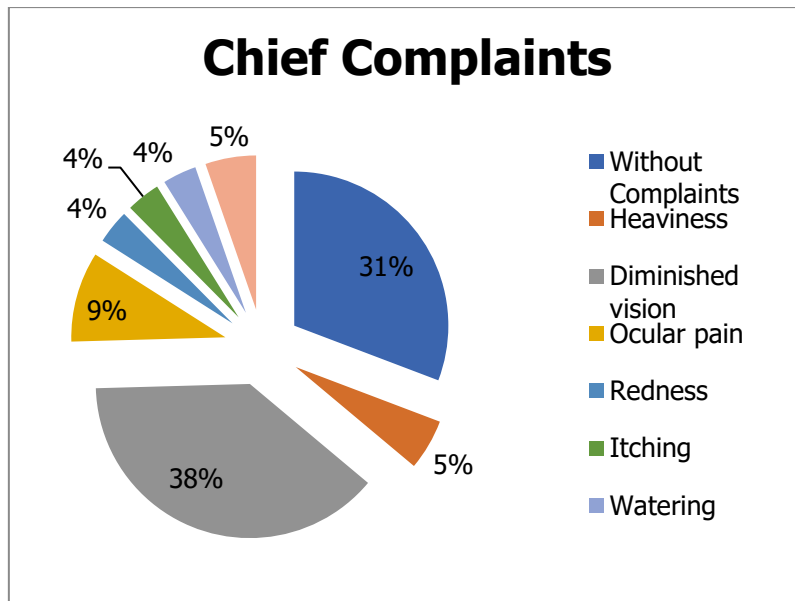


Fig:5

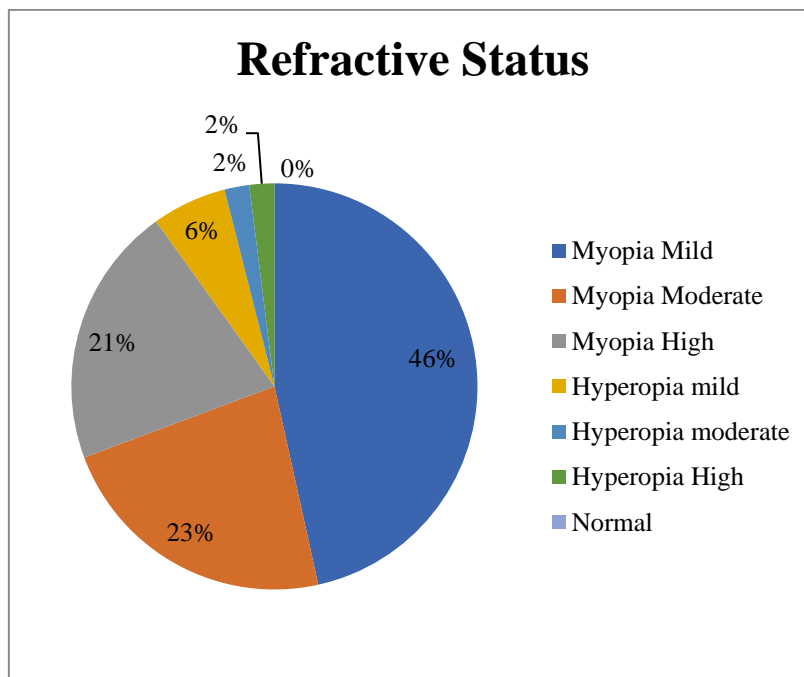


Fig 6

It was also found that 83% of eyes had no visual impairment, 10% were blind, 4% had severe visual impairment, whereas 3% had mild to moderate visual impairment. (fig 7).

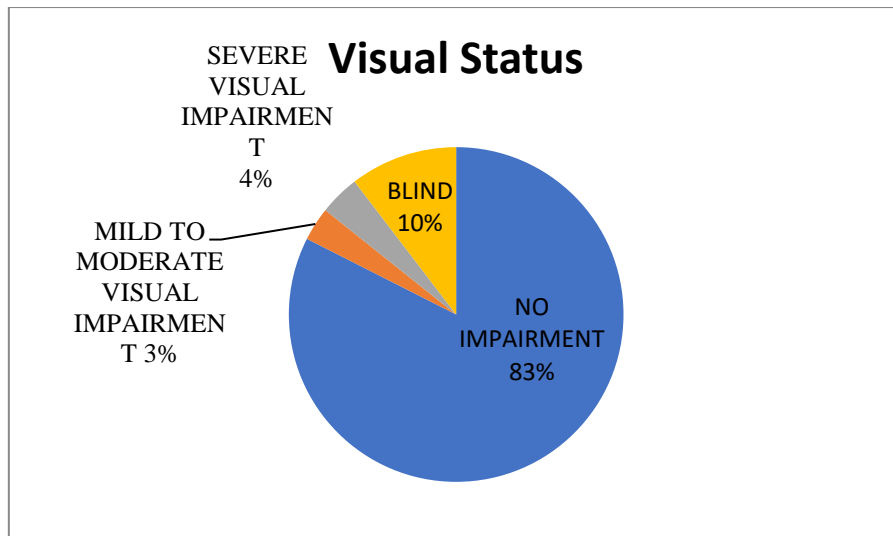


Fig 7

When looked for clinical features we found that 69% eyes had no specific features, 16 % were status post trabeculectomy, 6% had pupillary abnormalities, 4% had corneal abnormalities, 3% had conjunctival pathologies and 2% had iris abnormalities. (fig 8)

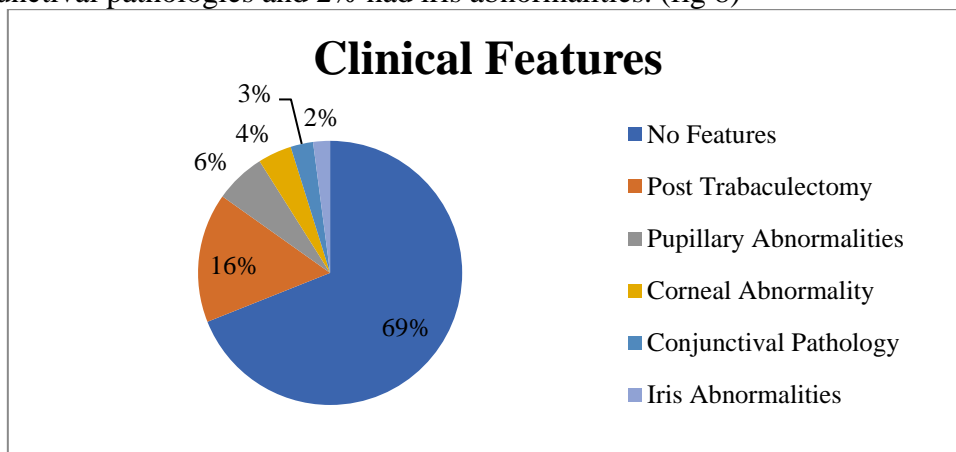


Fig: 8

42% eyes had advanced cupping, 35% had moderate cupping, 23% had normal CD ratio (fig: 9)

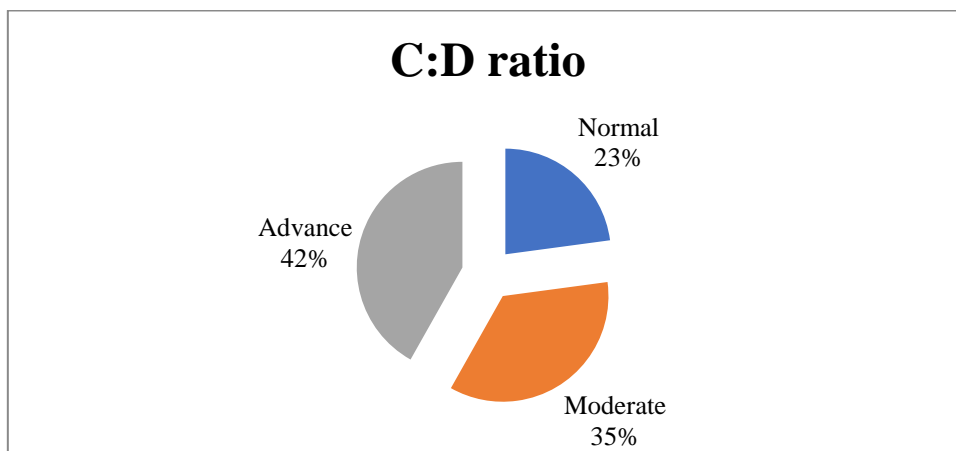


Fig 9

75 % eyes had a medium disc size, 21% had large disc and 4% had small disc size. (fig 10)

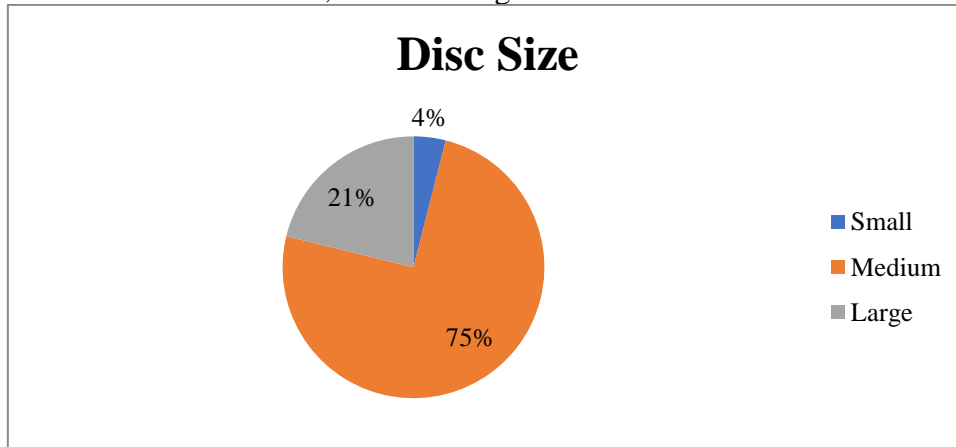


Fig 10

30% had advanced rim loss, 28% had superior thinning, 19 % had different disc anatomy, 13% had bipolar thinning, 5% had glaucomatous optic atrophy, 4% had disc pallor, 1% had inferior thinning. (fig 11)

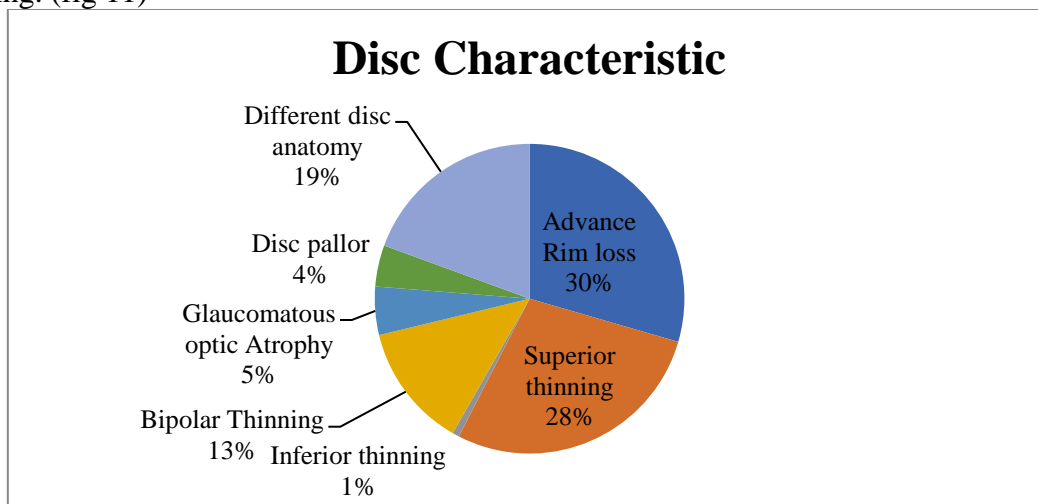


Fig 11

30% of eyes had no visual field defect, 23% had few depressed points, 21% had advanced field loss, 6% had biarcuate defect, 6% had inferior defect, 6 % had superior arcuate, and 8% had nasal step. (fig 12)

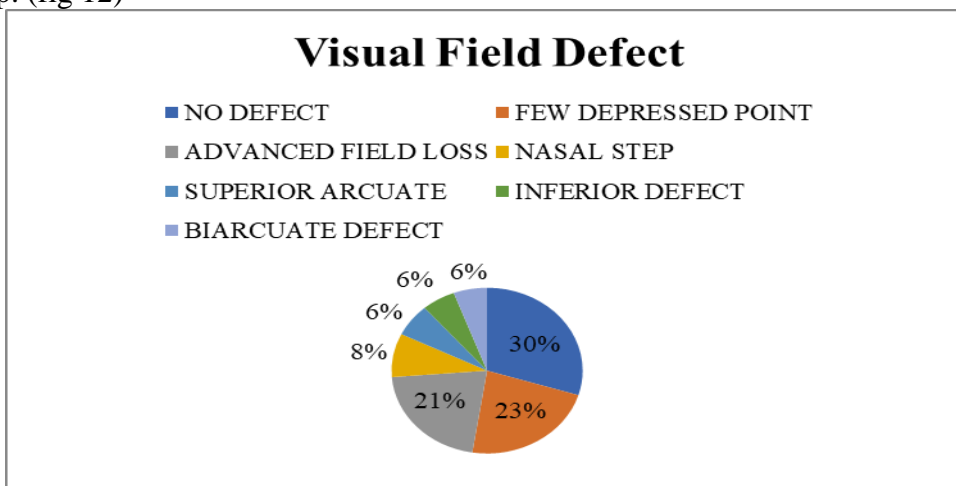


Fig 12

Out of 154 eyes, 131 patients were undergoing medical and surgical treatment. And the mean IOP was 19.2105 with a Range of (4-52mm Hg). Rest 23 patients were not undergoing any treatment (i.e., Surgery and Anti Glaucoma Medications), and the mean IOP was 19.133 with a range of (7-44mm Hg), shows the IOP profile among the patients reviewed.

DISCUSSION

In this study, we showed that JOAG patients were predominantly male, had myopic refractive states, and had severe IOP elevations; these findings were consistent with those of earlier studies [11-13]. Interestingly it was seen that a large amount of JOAG patients was under treatment (i.e. Surgery and Anti Glaucoma Medications) and the IOP range was about (4-52 mm Hg). Out of which 69.7 % of the patients were of Post Trabeculectomy. The patients with unilateral JOAG had higher chance of developing JOAG in their fellow normal eyes, similar findings were noted in a study done in Korea.^[14] More caution is needed during follow-up to identify these patients so that they can receive proper treatment. Symptoms mostly associated with diminished vision including heaviness and ocular pain. However, 33.6% of patients visited the hospital without any definite symptoms and was diagnosed with JOAG. In the visual field mean VFI was 74% and SD (0.304). A family history was weakly associated as there was a high percentage of patients (87%) who were not aware of their family history of glaucoma. Contrarily, Wu et al. [15] found that patients with familial POAG had a more severe disease and were diagnosed at a younger age than those with sporadic disease.

According to earlier research, the relationship between a family history of glaucoma and the development or severity of the VF defect in POAG patients was debatable. There haven't been any reports of this in JOAG patients, though. Additionally, 12.5% to 36% of cases of JOAG may have been related to myocilin mutations, while the

prevalence of a myocilin mutation in POAG patients ranged from 1.4% to 4.3% globally [16,17]. Therefore, it is possible that a family history in JOAG patients might be associated with VF progression.

Lastly it was seen that there was no specific clinical pathology and systemic illness among JOAG patients noted in this study.

This study was actually aimed to analyse the clinical characteristics and profile of the JOAG. So, we analysed according to the refractive error, cupping of disc, disc characteristics, clinical features and the Visual Field Defect. A similar study with other correlating factors like familial inheritance, RNFL and GCC loss and other systemic conditions with incorporating more patients or similar number of patients is advised for further analysis.

CONCLUSION

In conclusion, we confirmed that JOAG patients presented with a male preponderance, a myopic refractive state and severe elevation of IOP. Periodic eye examinations are needed because a considerable number of JOAG patients have no definite symptoms. These findings indicated that JOAG patients may require more careful management of IOP. The limitation to this study was first, there might have been selection bias and confounding factors due to the retrospective design. Drop-out during follow-up may have happened in cases of poorly treated patients. The clinical backgrounds, including the age, sex, and baseline IOP, did not, however, differ significantly. Due of its rarity, the second drawback was the relatively small overall patient population, which made it challenging to distinguish between group. Additional extensive prospective large-scale investigations would be required for a firm conclusion.

Conflict Of Interests: There were no declared conflicts of interest that would have affected this article.

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