

Screening of Accompanying First Degree Relatives of Patients with Primary Open Angle Glaucoma

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Abstract

Introduction: Glaucoma is one of the common causes of blindness worldwide, and the leading cause of irreversible blindness. First-degree relatives of POAG patients have 4–16% risk of developing POAG. Though poorly understood, primary open angle glaucoma (POAG) is believed to have a genetic or familial component that may occur through polygenic or multifactorial transmission. **Objective:** To assess the screening of accompanying first degree relatives of patients with primary open angle glaucoma. **Methods:** The authors performed a hospital-based cross-sectional study at a Department of Ophthalmology, Ad-din Akij Medical College, Khulna & Bangladesh Eye Hospital, Khulna, Bangladesh from July to December 2022. A total of 60 first degree relatives of POAG patients were included in the study. All first-degree family members of POAG patients who accompanied them to the glaucoma clinic underwent a full ophthalmic examination. The optic disc was evaluated and intraocular pressure (IOP) was measured. POAG and glaucoma suspect were defined as per ISGEO classification. **Results:** 60 first degree relatives of 52 glaucoma patients were examined. The mean age was 30.67 years (± 12.71). 54 (90.0%) of 60 of accompanying first degree relatives were off springs, 6 (10.0%) were siblings. 5 out of 60 (8.3%) were diagnosed as glaucoma and started on anti-glaucoma medication. 14 (23.3%) were glaucoma suspects. 41 (68.4%) of the participants had no sign of glaucoma. **Conclusion:** Ocular examination of the first-degree relatives accompanying POAG patients helped to identify a remarkable number of individuals with glaucoma and thus might be used as an effective and viable tool for screening glaucoma in a hospital setting. Awareness regarding glaucoma is very low even among the first degree relatives of glaucoma patients.

Keywords: Awareness, Glaucoma, POAG, Relatives.

Introduction

Glaucoma is one of the common causes of blindness worldwide, and the leading cause of irreversible blindness (1). First-degree relatives of POAG patients have 4–16% risk of developing POAG. Given the circumstances and the prevalence of POAG in first degree relatives of POAG patients, screening the first degree relative/s accompanying the patient to the hospital can be a cost effective and viable tool for glaucoma screening that will not add any extra cost of travel and accommodation. An estimated 57.5 million people worldwide are affected by open angle glaucoma with a global prevalence of 2.2% (2). Primary OAG is a chronic optic neuropathy, which occurs with an open angle in the absence of other explanatory causes. Primary open-angle glaucoma (POAG) has a genetic or familial component. It is believed that the genetic influence occurs through polygenic or multifactorial transmission. Reportedly, 5–50% of cases of POAG are hereditary, with the best estimate being 20–25%. The risk of developing POAG in first-degree relatives is 4–16% (3-5). The disease has a hereditary component and becomes more prevalent with age. POAG progresses very slowly and is usually asymptomatic until late in its course, so affected individuals can develop severe damage before they seek professional help. POAG has been shown to be more prevalent in first-degree relatives, so their screening for glaucoma is important. Various studies have reported different prevalence depending on the population sampled, the age of the individuals studied, the techniques of examination, and the definitions of glaucoma used. Although there is no standard definition, current criteria require the presence of visual field and optic disc damage, regardless of intraocular pressure (IOP) (6). Though poorly understood, POAG has genetic of familial component that may occur through

polygenic or multifactorial transmission (7, 8). Reportedly, 5–50% of cases of POAG are hereditary, with the best estimate being 20–25% and the risk of developing POAG in first-degree relatives is 4–16% (9-11). Likewise, the relative risk of developing POAG in a population with a positive family history is 9.2 (12). Many studies suggest that approximately 5% of POAG results from mutations in the myocilin (MYOC) gene, and thus individuals predisposed to the development of POAG can be identified to some extent (9,13,14). However routine genetic screening for the mutation is not feasible in a developing country like ours. Given the circumstances and the prevalence of POAG in first degree relatives of POAG patients, screening the first degree relatives can be a cost effective way of diagnosing the disease in its early stage. We performed this study to investigate whether screening the first degree relative accompanying the glaucoma patient to the hospital may be a viable tool for glaucoma screening.

Materials and Methods

The authors performed a hospital-based cross-sectional study at a Department of Ophthalmology, Ad-din Akij Medical College, Khulna & Bangladesh Eye Hospital, Khulna, Bangladesh from July to December 2022. A total of 60 first degree relatives of POAG patients were included in the study. All first-degree family members of POAG patients who accompanied them to the glaucoma clinic underwent a full ophthalmic examination. The optic disc was evaluated and intraocular pressure (IOP) was measured. POAG and glaucoma suspect were defined as per ISGEO classification. Data entry and statistical analysis was done using Statistical Package for the Social Sciences (SPSS version 21).

All the people who were accompanying POAG patients to the glaucoma department were asked about their relationship with the patients and only the first degree relatives were included in the study after their consent. Ophthalmic history was obtained from the participants who accompanied the glaucoma patients to the glaucoma clinic. They were asked if they had heard about glaucoma. An answer of “yes” indicated that the subject was “aware” of glaucoma. Participants were interviewed regarding past history of any ocular examination and any history of examination related to glaucoma and its treatment. They then underwent a full ophthalmologic examination. The study excluded relatives of patients with closed or narrow angles and secondary glaucoma. Slit lamp biomicroscopy with a Volk’s 90 D lens was used to examine the optic disc, and Goldmann applanation tonometry was used to assess the intraocular pressure (IOP). Cases with IOP under 21 mmHg without medication and absence of typical glaucomatous optic disc changes were classified as normal (N) and advised for annual follow up. Participants having IOP more than or equal to 21 mmHg in either eye, or CDR asymmetry of more than 0.2, or having CDR ≥ 0.6 without typical glaucomatous optic disc damage or visual field change were referred for visual field test (Sita Standard automated static perimetry 24-2 program on the Humphrey Visual Field Analyzer [Zeiss- Humphrey Systems, Dublin, CA]). Visual field results were analysed by three experienced Glaucoma specialist for the presence of any glaucomatous visual field changes. POAG and glaucoma suspect was defined as per ISGEO classification.

Results

A total of 60 first degree relatives of POAG patients were included in the study, among which 28 (46.6%) were female and 32 (53.4%) were male. The mean age of the cases was 30.67 ± 12.71 years. The mean age in case of females was 32.86 ± 14.52 years, while that in case of male was 28.82 ± 10.84 years. 54 (90.0%) out of 60 accompanying first degree relatives were off springs and 6 (10.0%) were siblings. Out of 60 participants only 20 (33.3%) had their eyes checked up in the past and among which 2 (3.3%) had their eyes evaluated for glaucoma and also had their IOP measured with GAT. None of the participants were diagnosed with cases of glaucoma and thus none of them were under any type of anti-glaucoma medication. Out of 60 participants only 8 (13.33%) were aware of glaucoma. Among all the participants, 5 (8.2%, 3 female and 2 male) cases had optic disc and visual field changes supportive of glaucomatous damage and were diagnosed as glaucoma and started on glaucoma medication. Out of these 5 cases, 3 were offsprings and 2 were siblings. 13 participants (21.6%, 6 female and 7 male) were diagnosed as glaucoma suspects, among which, 12 were offsprings. 40 participants (66.6%) had no signs of glaucoma.

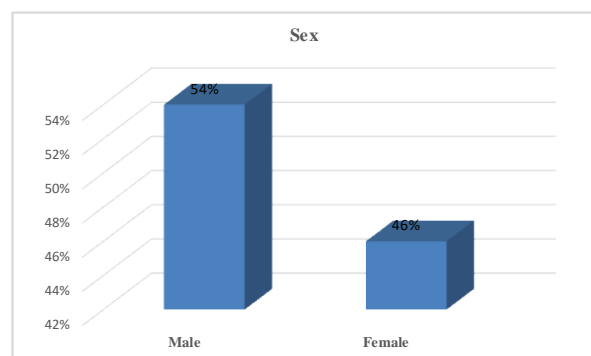


Fig-1: Sex distribution of the patients.

Table-1: Subjects already diagnosed as glaucoma and glaucoma suspects.

participants	N	%
FirstDegree	50	83.4%
Siblings	10	16.6%

Table-2: Glaucoma and glaucoma suspects their eyes checked.

IOP measured with GAT	N	%
Past IOP	20	33.3%
GAT	02	3.3%

Discussion

Family history is one of the important risk factors for the development of POAG [9, 10]. Since the visual field loss from glaucoma is irreversible, early detection and treatment is the only way to limit the damage from the disease. However, there is no proven feasible community based screening for detecting glaucoma established till date. Thus, this study was focused on screening a target population that comprised the first degree relatives of POAG patients who accompanied them to the glaucoma clinic. In our study, 8.25% of the participants had glaucoma, which is much higher than the prevalence (1.24%) of open angle glaucoma reported in the Bhaktapur glaucoma study Thapa et al. (15). This may be due to the inclusion of only first degree relatives of POAG patients in our study while the Bhaktapur glaucoma study was a population based study. The mean age of the participants in our study was 30.67 years ($\pm 12.71\%$), while that in the Bhaktapur glaucoma study was 55.4 ± 12.3 years. Despite their young age, the Barbados family study revealed that about a quarter of the relatives had OAG or suspected OAG Leske et al. (10). In our study, it was found that 35.8% of patients diagnosed with POAG before the age of 50 years had positive family history as compared to only 11.7% in patients older than 70 years (16). Findings from these studies are consistent with our finding, suggesting that screening first degree relatives might

help in early diagnosis of glaucoma. Vegini et al. (11) reported the prevalence of glaucoma in first degree relative as 16.8%. The Glaucoma inheritance study from Tasmania included 442 relatives of glaucoma patients and reported the prevalence as 18% McNaught et al (9). Likewise in another study Kong et al. (17) of 531 first-degree relatives, 67 (12.62%) were identified to have POAG, a rate eight times higher than that of the control group (8 of 526, 1.52%). All of these studies showed a higher prevalence of glaucoma than our study that may be attributed to the higher mean age of the participants in these studies; which was: 54 years McNaught et al. (9) 48.2 years old (± 11.15) Vegini et al. (11) and 58.53 (± 13.70) Kong et al. (17). In our study 23% of the participants were classified as glaucoma suspects, which is similar to the findings (30%) by Gupta P et al. (18). The higher prevalence of glaucoma suspects in first degree relatives suggests future risk of developing POAG in the participants. In our study among 5 (8.2%, 3 female and 2 male) cases had optic disc and visual field changes supportive of glaucomatous damage and were diagnosed as glaucoma and started on glaucoma medication. Despite accompanying a glaucoma patient to the glaucoma clinic, only 14.75% of the participants were aware of glaucoma in our study. Thus, relatives of glaucoma patients are more likely to be aware of glaucoma than the general population in Bangladesh, however, this percentage is still much lesser compared to the findings of other studies done in developed countries. Studies done in developed countries observed much higher awareness in relatives of glaucoma patients, 79% in the general population Livingston, McCarty and Taylor et al. (19) and 82% in the relatives Celebi AR et al. (20), which was attributed to higher levels of education. The lower observed awareness level in our study may be due to the

lower level of education in our society. The level of education however was not analysed in this study. This suggests that the family members of the glaucoma patients still lack awareness regarding glaucoma and the importance of glaucoma evaluation among them. Our study has few limitations. We included only accompanying first degree relatives, thus a large number of relatives might have been left out. We also did not study other risk factors except for being a first degree relative in the study. The reasons why not all first-degree relatives could be included were: migration of relatives to different cities, inability to come to the hospital for examination, and lack of interest for the examination. This again projects the difficulty in screening all first-degree relatives of glaucoma patients. Furthermore, we compared our results mostly with non-Asian countries, which may also account for a difference in prevalence rates. This shows the lack of awareness about increased hereditary risk of glaucoma among the general population. Although the subjects were aware of the fact that their relative had glaucoma, they did not know that they were at increased risk and needed to undergo glaucoma screening. Our study emphasizes the need for screening first-degree relatives of patients with POAG/NTG. Selective screening of first-degree relatives of POAG/NTG patients helps in early disease diagnosis. We also need a large population-based study to find out the prevalence rates of glaucoma in relatives of glaucoma patients among the Bangladeshi population.

Conclusion

Ocular examination of the first-degree relatives accompanying POAG patients helped to identify a significant percentage of individuals with glaucoma and thus might be used as an effective and viable tool for screen-

ing glaucoma in a hospital setting. Awareness regarding glaucoma is very low even among the first degree relatives of glaucoma patients.

Conflict of Interest: None.

Source of Fund: Nil.

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