Original Article

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 .The overall prevalence of glaucoma in Nepal is 1.9%. 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. 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.

Materials and methods: The authors performed a hospital-based cross-sectional study at a tertiary eye hospital in Nepal. 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: Sixty one first degree relatives of 54 glaucoma patients were examined. The mean age was 30.67 years (±12.71). Fifty five(90.2%) of 61 of accompanying first degree relatives were off springs, 6 (9.8%) were siblings. Five out of 61 (8.2%) were diagnosed as glaucoma and started on anti-glaucoma medication. Fourteen participants (23%) were glaucoma suspects. Forty two (68.9%) 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.

Key words: Awareness, Glaucoma, POAG, Relatives.

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INTRODUCTION

Glaucoma is one of the common causes of blindness worldwide, and the leading cause of irreversible blindness (Flaxman et al., 2017). An estimated 57.5 million people worldwide are affected by open angle glaucoma with a global prevalence of 2.2%(Wiggs and Pasquale, 2017).A population based study showed that the overall prevalence of glaucoma in Nepal was 1.9 %(Thapa et al., 2013). According to a survey, glaucoma was responsible for 5.9% of total bilateral blindness in Nepal(Y.D. Sapkota 2012).Primary OAG is a chronic optic neuropathy, which occurs with an open angle in the absence of other explanatory causes. Although there is no standard definition, current criteria require the presence of visual field and optic disc damage, regardless of intraocular pressure (IOP) (Foster et al., 2002). Though poorly understood, POAG has genetic of familial component that may occur through polygenic or multifactorial transmission (Lichter, 1994; Quigley, 1998).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% (McNaught et al., 2000; Leske et al., 2001; Vegini et al., 2008). Likewise, the relative risk of developing POAG in a population with a positive family history is 9.2 (Wu et al., 2006). 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 (Wiggs et al., 1998; McNaught et al., 2000;R. Frezzotti, A. Renieri, P. Frezzotti, 2004).

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. However, in a country like Nepal where affordability is a major barrier to easy accessibility of eye care, first degree relatives of any glaucoma patient may never visit distant clinics for glaucoma screening (Y.D Sapkota 2012). Thus, every effort can be made to examine any relative visiting the hospital with the patient as this will not add any extra cost of travel and accommodation. 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 tertiary eye hospital in Nepal from30thMarch 2020 to 1stMarch 2021. The study followed the principles of the Declaration of Helsinki. The study was approved by the Tilganga Institute of Ophthalmology (TIO) Institutional Review Committee in Kathmandu, Nepal, and all subjects gave written informed consent. Data entry and statistical analysis was done using Statistical Package for the Social Sciences (SPSS version 20.0).

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. Only the first-degree relatives irrespective of their age who were accompanying a POAG patient to the glaucoma clinic at Tilganga Institute of Ophthalmology, Kathmandu, Nepal were included in the study. 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 61 first degree relatives of POAG patients were included in the study,among which 28 (45.9%) were female and 33 (54.1%) were male. The mean age of the cases was 30.67 ±12.71years. 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. Majority of the cases were Mongolian (22; 36.1%), followed by Chettri (12; 19.7%), Newar(10; 16.4%), Bhramin(8; 13.1%) and others (9; 14.8%). Fifty five (90.2%) out of 61 accompanying first degree relatives were off springs and 6 (9.8%) were siblings.

Out of 61 participants only 21 (34.4%) 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 61 participants only 9 (14.75%) 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. Fourteen participants (23%, 6 female and 8 male) were diagnosed as glaucoma suspects, among which,12 were offsprings. among Forty two (68.9%) participants had no signs of glaucoma.

DISCUSSION

Family history is one of the important risk factors for the development of POAG(McNaught et al., 2000; Leske et al., 2001).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, 8.2% 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., 2013). 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 (±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., 2001). In one 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 (Budde and Jonas, 1999). 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 (2008) 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., 2000). Likewise in another study (Kong et al., 2013)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., 2000);48.2 years old (±11.15)(Vegini et al., 2008) and58.53 (±13.70) (Kong et al., 2013).

In our study 23% of the participants were classified as glaucoma suspects, which is similar to the findings (30%)by Gupta P.(Gupta and H, 2018). The higher prevalence of glaucoma suspects in first degree relatives suggests future risk of developing POAG in the participants.

Despite accompanying a glaucoma patient to the glaucoma clinic, only 14.75 % of the participants were aware of glaucoma in our study.Though 14.75% is a small percentage, it is still 6 times more than that found in the Bhaktapur glaucoma study (2.4% in general population)(Thapa et al., 2013). Thus, relatives of glaucoma patients are more likely to be aware of glaucoma than the general population in Nepal, 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, 1998) and 82% in the relatives(Celebi A.R., 2018), 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.

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

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