ORIGINAL ARTICLE Prevalence of Glaucoma in First Degree Relatives of Known Glaucoma Patients

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ABSTRACT

Objective: To determine the prevalence of glaucoma in first degree relatives of known glaucoma patients.

Methods: An observational, descriptive, cross-sectional study was carried out to determine the clinical and epidemiological behavior of primary open angle glaucoma (POAG) in 377 relatives of glaucomatous patients, of whom 195 (52%) were suspect to POAG. The patients who were already diagnosed with glaucoma and they came to eye ward for their follow up checkups, and those who were admitted for glaucoma surgery, their blood relatives were included in the study. We included those members of a family who voluntarily presented themselves for the research following inclusion & exclusion criteria. The data collected was statistically analyzed and the results were tabulated.

Results: Out of 377 participants 51% were male, majority of cases were between 40-60 years of age, three types of Glaucoma were prevalent in these individuals, 60% of individuals were having Primary Open Angle Glaucoma, 25% were having Ocular Hypertension glaucoma and 15% were having Normal Tension Glaucoma. The POAG was diagnosed in overall 45% of male and 40% of female population. **Conclusion:** According to the results, the prevalence of the POAG is much higher in individuals having family history with Glaucoma. In addition, investigations of all adult subjects from families with POAG yields new cases. The high prevalence of POAG among individuals having positive family history clearly indicates that strong relationship between genetics and Glaucoma.

Key Words: Glaucoma, POAG, First-degree Relatives.

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INTRODUCTION:

Among different causes of blindness, glaucoma is the second most common cause and is a significant cause of permanent visual loss in the world¹. According to the WHO around 13% of the 37 million blind people globally are affected with glaucoma. It has been envisaged that occurrence of glaucoma will certainly rise to 80 million with an estimated 11 million persons bilaterally blind from glaucoma by the year 2020².

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<u>Correspondence to:</u> Dr. Kainat Saleem Women Medical Officer, Ophthalmology Deptt. PUMHSW-SBA Email: drkainat.saleem@gmail.com The increased risk of occurring glaucoma in family members of Glaucoma patients has been well-recognized and several studies have reported on screening of relatives of individuals with open angle glaucoma³.

A study conducted in United Kingdom shows that 75% of Asian children (Indian, Pakistani, and Bangladeshi) had primary glaucoma compared with 33% to 43% of children of other ethnic origins⁴. In a study in Rawalpindi Pakistan, 87.5% of the subjects with normal intraocular pressure were found to have suspicious glaucomatous cupping⁵. A study conducted in ophthalmology department of Postgraduate Medical institute Peshawar shows that 40% of the participants were blind due to glaucoma⁶.

It has long been known that children of glaucoma patients have a higher likelihood of developing glaucoma⁷. There is no doubt that regular eye examination should be recommended for all family members of glaucoma patients.

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If glaucoma developed during early childhood, then all relatives both young and old should be examined⁸. If glaucoma occurred at a later age, then examination of adult relatives is sufficient. Those with a family history of glaucoma should have their eyes examined between ages of 20 and 30 but certainly no later than 30 to 40⁹. A comprehensive screening of families of diagnosed cases of Glaucoma can prevent the blindness and thus improve the quality of life in community.

In India Juvenile-onset open angle glaucoma (JOAG) patients with family history of glaucoma tend to present earlier and to with less severe disease, probably because of early detection¹⁰. This shows that early detection of glaucoma results in better visual outcomes.

A study in Shanghai shows that relatives of glaucoma patients have a strong risk of developing glaucoma. Out of 531 first-degree relatives, 67 (12.62%) were identified to have POAG⁹. Siblings of the patients suffering from primary angle closure glaucoma (PACG) are at elevated risk for the condition, a two third of them shows clinical findings related to glaucoma, suggested by a study¹⁰ in which 47 families 95 siblings with at least one person affected with glaucoma took participation in the research; 58% of the total individuals were classified within the scale of angle closure, 9.5% patients with open angles showed other anomalous characteristics, within these individuals 5.3% cases of primary openangle glaucoma, 3.2% subjects with suspicious discs, and 1.1% with normal-tension glaucoma. The 32.6% had no evidence of glaucoma. It is suggestive that if any person in the family is affected with primary angle closure glaucoma, the other family members can be at risk of any type of primary glaucoma including primary open angle glaucoma and normal tension glaucoma.

A study¹¹ conducted in China shows that a family history of glaucoma is associated with the presence and severity of primary angle closure glaucoma and primary open angle glaucoma. Of the 332 primary angle closure glaucoma patients, 83 (25.00%) had glaucoma family history. Of the 228 primary open angle glaucoma patients, 49 (21.49%) had a family history of glaucoma. This supports the finding that screening the first-degree relatives will be an effective way to detect any type of primary glaucoma in a population¹².

The ongoing researches in the fields of glaucoma are aimed at the early diagnosis of glaucoma in the people who are at a high risk of developing the glaucoma in order to prevent blindness.

METHODS:

The research study was conducted at the department of Ophthalmology, Peoples Medical College Hospital Nawabshah. The research was based on cross sectional study based on convenience sampling technique, 377 individuals from different families were included in the research process. The blood relatives of diagnosed cases of glaucoma, attending the eye ward for their follow up checkups, and those who were admitted for glaucoma surgery, were included in the study. We included those members of a family who voluntarily presented themselves for the research following inclusion & exclusion criteria. *The following criteria were taken into account:*

Inclusion criteria: Relatives of first degree of relationship, age: 20 years or more, transparency of the refractive media capable of allowing the study of the fundus. Exclusion criteria: Family or second degree of kinship, age: younger than 20 years who accept not be part of this research, with pathologies that obstruct the view of the details of the fundus. Patient suspected: Family history of POAG in a first - degree relative, intra ocular pressure (IOP) of 24 to 27 mmHg without defects in the retinal nerve fiber layer (RNFL), excavation ratio - greater than 0.7 papilla, asymmetry of greater than or equal to 0.2 papilla, normally.

Glaucomatous patient: Home in adulthood, IOP greater than 21 mmHg at some point in the course of the disease, open angle normal looking head injury glaucomatous NO, visual field loss. Finally, glaucomatous patients were classified according to clinical stages of the disease.

The data was analyzed by SPSS version 22 to see the prevalence of glaucoma in accordance with age, gender and relation with the glaucoma patient. The continuous variable like age was analyzed by frequency mean and standard deviation while gender, relation with the patient, type of glaucoma in patient, diagnosis of glaucoma in participant and type of glaucoma in participant were analyzed by frequency.

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Gender	Frequency	%	
Female	184	49	
Male	193	51	

Table I. Frequencies of Gender Representation

Participants with different ages were included in the research in order to make sure diversity in the research. According to the research, it was found that Glaucoma is mostly found in people above 40 years of age. Therefore, the respondents from 20 age onwards were selected. 110 participants (29%) were in age group 20-40 years, 160 participants (42%) were in age group 40-60 years, 77 participants (20%) were in age range of 60-80 years and 30 participants (8%) were above 80 years of age (table II).

Table II. Frequencies Age Range

Age Range Frequencies	Individuals	
20 - 40	110 (20%)	
40 - 60	160 (42%)	
60 - 80	77 (20%)	
Above 80	30 (8%)	

According to the results three types of Glaucoma were prevalent in these individuals such as Ocular hypertension (OH), Primary Open Angle Glaucoma and Normal Tension Glaucoma. The findings of the research revealed that 60% individuals were having Primary Open Angle Glaucoma, 25% were having Ocular Hypertension glaucoma and 15% were having Normal Tension Glaucoma (chart I)

Type of Glaucoma Diagnosed in Individuals



Chart I: Patients with Glaucoma Type

In addition, individuals from different age groups were diagnosed glaucoma. According to the results, in the age group 20 to 40, 30% individuals were having POAG, 50% were having OHG and 20% individuals were having NTG. Similarly, in age group from 40 to 60, 40% individuals were having POAG, 25% were having OHG and 35% were having NTG. Finally, the results from age group 60 to 80 show that 30% individuals were having POAG, 25% were having OHG and 45% were having NTH (table III).

Table III. Age Ratio in Individuals having Different Tyes of Glaucoma

20 to 40		40 to 60			60 to 80			
POAG	OHG	NTG	POAG	OHG	NTG	POAG	OHG	NTG
30%	50%	20%	40%	25%	35%	30%	25%	45%

Likewise, the table below shows the ratio of gender in individuals having different types of Glaucoma.

Table IV. Gender Ratio in Individuals having Different Types of Glaucoma

Male			Female		
POAG	OHG	NTG	POAG	OHG	NTG
45%	35%	20%	40%	25%	35%

DISCUSSION:

Glaucoma is a condition of the eye characterized by progressive deterioration of the optic nerve. In most cases it is a chronic illness and can lead to an incapacitating loss of vision¹³. Optic nerve injury is manifested by an excavation of the optic disc with loss of nerve fibers in the retina. A high Intraocular Pressure (IOP) is an important risk factor for the development of glaucoma. In the eye, the level of aqueous humor production, resistance in the drainage or outlet pathways, and the pressure of the episcleral veins, regulate the daily variations (day and night) of the IOP¹⁴.

Primary open-angle glaucoma accounts for 90% of all cases of glaucoma among the Caucasian population. In this type of glaucoma, the obstruction is due to partial and restricted outflow of the aqueous humor in the area of the trabecular tissue, is accepted as the cause that results in IOP increase. The process here is not as dramatic as in increase.

The process here is not as dramatic as in the case of angle-closure glaucoma, but over time it can affect vision causing blindness. It has been pointed out that ischemia of the optic nerve and / or mechanical deterioration of the Optic nerve may be a consequence of elevated intraocular pressure¹⁵. Primary open-angle glaucoma is an asymptomatic disease. Initially the peripheral visual field is gradually affected, leaving the central field of vision seemingly without any damage. Therefore, the visual acuity is not significantly affected until the more advanced stages of the disease. The diagnosis of open-angle primary glaucoma is perfc ned by measuring the IOP (tonometry)¹⁶, performing a visual field examination (perimetry), and examining the optic nerve head (ophthalmoscopy).

Epidemiologic findings reveal that among different risk factors, family history with glaucoma is the highest. It is indicated that more than half of patients with POAG have presence of glaucoma in their family¹⁷. Early diagnosis of glaucoma is important to delay the damage of the diseases and in curtailing permanent loss of vision¹⁸. The analysis of the researches revealed that a family history with glaucoma in 1st-degree relatives and screenings measures an IOP greater than 21 mm Hg. This value shows that the IOP have significant relationship with the growth of glaucoma¹⁹. A blend of these results presented a general sensitivity of 81.7% with a specificity of 55%²⁰.

In another family aggregation study, the glaucoma prevalence was more than 10.4% in the siblings of individuals, 1.1% in the offspring²¹. The lifetime risk of glaucoma was 22% in relatives of patients with glaucoma, almost ten times higher than in controls, suggesting that at least one sixth of all glaucoma cases in the general population may be caused by a genetic component²².

The results coincides with most of these studies and indicate that the prevalence rate of PAOG in individuals having positive family history is high. The tests were conducted by evaluating IOP defects in Corresponding Region, Optic disc defects / loss, Cup disc defect/loss and visual field defect. The findings of all these variables show that a high risk prevails in individuals having positive family history with glaucoma. In addition, the results further indicated the majority of the people having positive family history are unaware about the disease.

So, it signifies the importance of glaucoma screening particularly among the population with a positive family history of glaucoma.

The study had certain limitations which come across while investigating individuals. Many of these participants were not agree for a thorough eye checkup because according to them they were alright and did not need this examination, so it was a challenge for us to explain the scenario and due to this we had to omit a very important examination Gonioscopy, as the participants were not giving consent for the procedure. We made our diagnosis of open angle glaucoma on the basis of depth of anterior chamber by Van Herick method.

CONCLUSION:

According to the results, the prevalence of the POAG is much higher in individuals having family history with Glaucoma. In addition, this investigation of all adult subjects from families with POAG yields new cases. Even in large pedigrees with POAG, majority of them were not aware about their positive family history. The results further suggest that an increased percentage of POAG prevalence in individuals having positive family history with Glaucoma might be inherited. The research findings carry a lot of potential in the field of ophthalmology. Due to its focus on determining the prevalence of POAG in individuals having history in first degree relatives, the research carries a lot of significance. The research study is the blend of genetics and ophthalmology. As a result, the research findings have opened new dimensions in both fields.

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