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ORIGINAL ARTICLE

Knowledge and attitudes regarding etiology and genetic counseling among Saudi children with primary congenital glaucoma

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ABSTRACT

Background: Primary prevention of primary congenital glaucoma (PCG) includes improving families of children with PGC. We evaluated the level of knowledge and attitudes of parents of children on PCG in Saudi Arabia.

Methodology: This was a personal interview-based survey of parents of children with PCG at a tertiary eye hospital in Saudi Arabia. The study was conducted in 2018. A close-ended questionnaire in Arabic was used. Demographic data were collected on the children and parents. Clinical data on PCG were collected from electronic case records. Five questions each on knowledge and attitudes toward genetic counseling were queried. A Likert-type scale was used to collect the responses. Rasch analysis was carried out for knowledge and attitudes. The score was correlated with demographics and clinical determinants. $p < 0.05$ was considered statistically significant.

Results: The study sample comprised 60 participants. The median Rasch score for knowledge on genetic counseling for PCG was -4.57 [interquartile range (IQR) -7.28; -1.0]. The median Rasch score for attitudes toward genetic counseling for PCG was -8.9 (IQR -11.6: -5.9). Parents with more than one family member with PCG had a significantly higher knowledge than those with one family member with PCG ($p = 0.007$). Knowledge of etiology and genetic counseling was significantly better if the child had residual vision amenable to low vision care ($p < 0.001$). The Rasch scores for knowledge and attitude were positively correlated ($p < 0.001$).

Conclusion: Knowledge of the cause of PCG and genetic counseling was high among parents. The positive attitude toward genetic counseling could be useful for the primary prevention of CG in Saudi Arabia.

Keywords: Primary congenital glaucoma, genetic counseling, prevention, etiology, birth defects.

Introduction

Primary congenital glaucoma (PCG) is a significant cause of childhood blindness in the Kingdom of Saudi Arabia (KSA) (1,2). Al-Anazi et al. (3) estimated the prevalence of PCG in KSA as 1:3030 live births. In KSA, most PCG are autosomal recessive, with mutations in the CYP1B1 responsible for over 85% of the cases. KSA is a conservative society and the inheritance pattern and single-gene etiology provide an opportunity for genetic testing and counseling to reduce the incidence of the disease. Screening and genetic counseling programs have a positive impact on the prevention of inherited diseases. These programs identify individuals at risk and provide education about their health and the risk of having an affected offspring (4). Several countries have carried out obligatory screening programs for common genetic diseases. These initiatives resulted in a significant decline with up to 100% reduction in births of common hemoglobinopathies in Cyprus, Italy, and Greece (5). In Saudi Arabia, Memish et al. (6) studied the outcome of the national premarital screening and genetic counseling program for common hemoglobinopathies for over 6 years and found a significant decrease in at-risk marriages. These successful programs have encouraged eye care professionals to eradicate PCG, thereby significantly reducing the societal, financial, and psychological burden of childhood blindness (7). The cooperation of parents and guardians of children with genetic childhood disease

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is crucial for timely counseling and genetic testing (8,9). A study found that the level of knowledge of breast cancer's genetic etiology was low among adults, but interest in genetic testing was high (10). A study of a rural population with minimal knowledge of genetic counseling reported a positive attitude toward genetic testing and counseling (11). To the best of our knowledge, attitudes, and knowledge of genetic testing and counseling in KSA has not been studied. We present the level of knowledge and attitudes toward personal counseling and genetic testing among parents of children with PCG.

Subjects and Methods

The Institutional Research Board approved this study (1669-P). A questionnaire-based survey was conducted in 2018. Caregivers/parents of children with PCG were enrolled for this study. Subjects were recruited from glaucoma clinics and the waiting area of operating rooms at our institution. A PCG registry was also used to identify potential participants. Written informed consent was obtained for participation in the study. Those declining to participate were not included in the survey. To calculate the sample size for this cross-sectional study, we assumed that among 2000 children with PCG treated at KKESH, 45% of their parents would have an acceptable level of PCG knowledge. To achieve a 95% confidence interval (CI) and a 15% margin of error with a 1.5 clustering factor, 60 parents of 60 children had to be randomly selected and interviewed.

Demographic data were collected, including the number of children in the family, other children with PCG, the participant and spouse's education level, and consanguinity between spouses. In the survey instrument, five questions were related to knowledge about genetics, counseling, and risk factors for PCG. Another five questions were related to the attitude of the participant toward genetic counseling and testing (Appendix 1). A 5-point graded response on a Likert scale was used for each question as follows (12): a logit value of -2.63 for "strongly disagree"; -1.1 for "disagree"; 0 for a neutral response; 0.09 for "agree"; and +2.86 for "strongly agree". The sum of the logit scores for knowledge and sum of the responses for attitude was calculated to represent the overall knowledge and attitude of each participant (13). The knowledge and attitude score was correlated to demographics and PCG-related variables. The Mann-Whitney U test or Kruskal-Wallis tests as applicable were performed, with $p < 0.05$ indicating statistical significance.

Results

The study sample comprised the parents of 60 children with PCG. The children's median age was 3 years (25% quartile 1 year, minimum 1 year, and maximum 17 years). The profile of parents of children with PCG (participants) is given in Table 1. The participants had a high level of education and high consanguinity (Table 1). Approximately 10% of participants had another child with PCG. One in eight parents had more than one child with PCG in the extended family.

The median Rasch score for participant knowledge on genetic counseling for PCG was -4.57 (interquartile range -7.28; -1.0). The median Rasch score for participant attitude on genetic counseling for PCG was -8.9 (IQR -11.6: -5.9) (Figure 1).

Table 2 presents the knowledge score and its association with the determinants. The level of knowledge was significantly higher ($p = 0.007$) among parents with more than one family member with PCG than those with only one child with PCG. Table 3 presents the score for attitude and its association with determinants. Although a positive attitude score differed in subgroups of determinants, the outcome was influenced by the small sample in subgroups. Table 4 presents the correlation of parent knowledge on genetic counseling for PCG in relation to PCG's clinical presentation. The knowledge on etiology and genetic counseling among parents was statistically significantly better if their child had vision between 20/200 and 20/400

Table 1. *Profile of parents of children with congenital glaucoma surveyed for knowledge and attitude for genetic counseling in Saudi Arabia.*

		Number	Percentage
Number of children	2 or less	19	31.7
	3 and more	41	68.3
Other child with PCG in family	Yes	11	18.3
	No	49	81.7
Other child with PCG in extended family	Yes	14	23.3
	No	46	76.7
Participants education	School and less	15	25.0
	College and more	45	75.0
Spouse's education	School and less	14	23.3
	College and more	46	76.7
Consanguinity among parents of child with PCG	No	17	28.3
	First cousin	33	55.0
	Second cousin	9	15.0
	Other	1	1.7

PCG denotes primary congenital glaucoma.

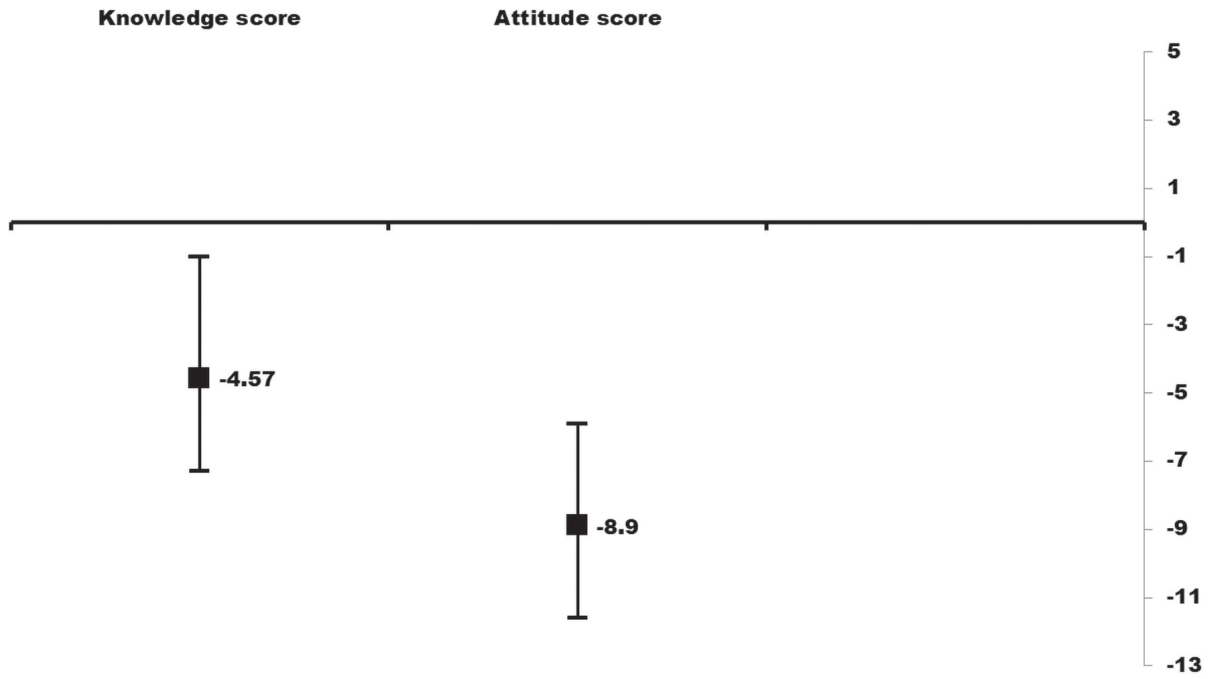

Figure 1. *Knowledge and attitude score regarding etiology and genetic counseling among parents of Saudi children with primary congenital glaucoma. (X-axis denotes knowledge and attitude for genetic counseling among parents and Y-axis denoted the Rasch score).*

Table 2. *Subgroups of awareness regarding common and blinding eye diseases among Makkah residents, Saudi Arabia.*

Subgroup of awareness	Median	Interquartile range
Sunglass and refractive error	0.2	0.0; 0.4
Contact lens	-0.5	-0.5; -0.25
Childhood blindness	-0.5	-0.75; -0.25
Eye diseases in old age	0.4	0.2; 0.4
Eye medication usage	0.0	0.0; 0.5
Eye screening	0.0	0.0; 0.0
Eye injury	0.5	0.0; 1.0

Variation in awareness among subgroup of eye component was significant (Friedman $p < 0.001$).

(residual vision amenable to low vision care) ($p < 0.001$). The globe's size was noted in 36 (60%) children (18 large, 2 small, and 16 normal-sized globes). Hence, we did not correlate this clinical parameter to the level of knowledge and attitude. Congenital cataract was present in two eyes. None of the children were using low vision aids. The Rasch score for knowledge and attitude was positively correlated ($p < 0.001$).

Discussion

This study provides unique insight into the knowledge and attitudes among parents of children with congenital glaucoma in KSA, where the disease's incidence is high.

The literature on the knowledge and attitudes regarding genetic testing for inherited ocular diseases is limited to a few publications on inherited retinal diseases (1,2) and some on retinoblastoma (14,15). The outcomes of the current study indicate that parental levels of knowledge were low, but acceptance/attitudes toward genetic testing/counseling were high. In KSA and other gulf countries, where premarital genetic testing is mandatory for several hemoglobinopathies, studies have demonstrated mixed results (16-19). A study from Jeddah, KSA, reported that the knowledge levels were low, but the attitude toward testing was very positive (16). The previous study's findings appeared to be associated with ethnicity, higher education, and higher income (6). On the contrary, a large cross-sectional study from Qatar (17) and a Syrian study with a smaller sample size (18) reported low levels of knowledge and a negative attitude toward genetic testing. Similarly, mixed results have been reported from the rural US Midwest (19). Our study found a high level of knowledge and an excellent attitude toward genetic testing and counseling among parents of children with PCG. There are no publications on the attitudes and knowledge related to genetic counseling for PCG, precluding comparison with published literature. In our study, both the knowledge and attitude toward genetic testing were highly positive. These factors could have been influenced by the fact that these patients had received treatment at an eye hospital where concomitant health education was often provided. Additionally, most parents were educated and had many immediate families (and likely, distant family members) with PCG. Hence, it is likely that they researched information on the internet to improve their knowledge of the condition. Furthermore, in this tertiary hospital, there is a reasonable level of patient trust for the physician. It is possible that this factor influenced their attitude toward genetic testing.

Table 3. *Attitude score for genetic counseling and its determinants among parents of Saudi children with primary congenital glaucoma.*

		Rasch score for attitude toward genetic counseling for primary congenital glaucoma				
		Median	**25% quartile**	**Minimum**	**Maximum**	**Validation**
Age						
Other child with PCG	Yes No	-10.1 -8.6	-11.6 -11.2	-13.2 -13.1	5.7 4.6	0.9
Other family member	Yes No	-9.5 -8.6	-11.6 -11.6	-13.2 -13.1	5.7 4.6	0.36
Consanguinity in parents	Yes No	-8.9 -7.0	-11.6 -10.1	-13.2 -13.1	5.7 4.6	0.34
Education of participant	School Higher	-6.1 -8.6	-11.6 -11.6	-13.1 -13.1	4.6 5.7	0.24
Education of spouse	School Higher	-7.8 -8.6	-11.6 -11.6	-13.2 -13.2	4.6 5.7	0.6

PCG denotes primary congenital glaucoma.

Table 4. *Correlation of knowledge score of genetic counseling among parents of children with primary congenital glaucoma to clinical presentation of glaucoma in their wards.*

		Rasch score for knowledge about genetic counseling for primary congenital glaucoma				
		Median	**25% quartile**	**Minimum**	**Maximum**	**Validation**
Age						
Laterality	Unilateral Bilateral	-7.8 -1.88	-8.6 -5.3	-8.6 -10.5	0.84 7.6	MW $p = 0.6$
Type of glaucoma	PCG Other	-2.5 0.27	-5.6 -7.7	-10.5 -8.6	7.6 4.7	MW $p = 0.2$
Vision in the better eye	20/20 to 20/60 <20/60 to 20/200 <20/200 to 20/400 <20/400 to PL	-4.6 -4.6 -10.3 4.1	-8.6 -7.3 -10.3 -4.4	-10.5 -9.0 - -10.1	2.4 4.7 - 7.6	Friedman $p < 0.001$

PCG denotes primary congenital glaucoma; PL denotes preferential looking; MW denotes Mann-Whitney; $p < 0.05$ is statistically significant.

Declaration of conflicting interests
The authors of this article have no affiliations with or involvement in any organization or entity with any financial interest or non-financial interest in the subject matter or materials discussed in this manuscript.

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Ethical Approval
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Consent for publication
Informed consent was obtained from the patients.

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