Parental experience with an ocular genetic counseling services in Saudi Arabia

Areej Alizary[1], Khabir Ahmad[2], Amani Al Bakri[3]

Access this article online

Quick Response Code:

Website: www.saudijophthalmol.org

DOI: 10.4103/sjopt.sjopt_154_23

Abstract:

PURPOSE: This study reports parental attitudes towards preimplantation genetic diagnosis (PGD) and their satisfaction with genetic counseling services offered by a territory eye care hospital in Saudi Arabia.

METHODS: This was a cross-sectional study of 30 parents (either father or mother) of children affected by recessive genetic ocular disorders. Their basic knowledge of recessive genetic disorders, attitude toward PGD, and satisfaction with counseling services were assessed using structured telephonic interviews.

RESULTS: Eighty percent of the participants understood the concept of recessive inheritance patterns and the effect of consanguineous marriages on raising the likelihood of giving birth to a child with genetic disorders. Forty-seven percent of parents understood the risk of having an affected future offspring. Sixty-seven percent of them mentioned that they would consider using preventive measures when conceiving next time. Seventy-three percent of participants stated that they would share the genetic test results with family members. Ninety percent of the parents were satisfied with the genetic counselor's ability to listen to them and the way the counselor explained the genetic information. There was a statistically significant association between parental willingness to share genetic test results with relatives and being given the opportunity to share information with their genetic counselor ($P = 0.01$). There was no association between the parental's knowledge and their willingness to consider using preventative measures in the next pregnancy.

CONCLUSION: Most parents had a basic understanding of recessive disease. However, they often struggled to comprehend the science and mathematical probabilities determining the recurrence risk in future pregnancies. This complexity makes it difficult for them to recall the information. Attitude toward PGD did not seem to be related with parent knowledge. A significant proportion of the parents expressed willingness to share their genetic test results with relatives, but some were hesitant due to fears of stigma. Most parents were satisfied with genetic counseling services.

Keywords:

Attitude, genetic counseling, ocular disorders

Introduction

The human eye is a structurally, functionally, and genetically complex sensory organ.[1] It has multiple segments including the anterior chamber and posterior chamber and contains highly specialized organs such as the retina, iris and ciliary body.[1]

Each of these segments could be affected by inherited, acquired, or multifactorial pathologies.[1] Many of these inherited eye disorders manifest from birth, while others have a late onset. Common inherited ocular disorders include glaucoma, congenital cataracts, retinal dystrophy, malformations, strabismus, and color deficiency.[2]

Genetic counseling delivers advice to individuals and families affected or at risk of hereditary disorders. It aims to increase the awareness of genetic diseases, to provide options regarding disease management, and to educate regarding recurrence risk and benefits of further testing.[3] Genetic counseling can play a vital role in reducing the prevalence of inherited disorders and provides an understanding of the heredity

[1]Ocular Genetic Services, King Khaled Eye Specialist Hospital, Alfaisal University, [2]Ophthalmic Epidemiology, King Khaled Specialist Eye Hospital, [3]Department of Ophthalmology, King Fahad Medical City Riyadh, Saudi Arabia

Address for correspondence:
Ms. Areej Alizary, King Khaled Eye Specialist Hospital, P.O. Box: 7191, Riyadh 11462, Saudi Arabia.
E-mail: areej.Alizary@gmail.com

Submitted: 11-Jul-2023
Revised: 31-Jul-2023
Accepted: 05-Aug-2023
Published: 24-Oct-2023

How to cite this article: Alizary A, Ahmad K, Al Bakri A. Parental experience with an ocular genetic counseling services in Saudi Arabia. Saudi J Ophthalmol 2023;37:296-300.

aspect of the disease. It can help the participant make important life decisions that will benefit the entire family.

Numerous studies have assessed the effectiveness of genetic counseling. To our knowledge, this is one of the first studies to investigate parents' experiences with an ocular genetic counseling service for their children with hereditary eye disorders in the Kingdom of Saudi Arabia.

1. Investigate whether there is an association between the parent's knowledge and their willingness to consider using preventative measures in the next pregnancy
2. Determine the association between parent's satisfaction with their counseling sessions and their willingness to share the results of genetic testing with their relatives
3. Examine whether there is an association between the attitudes of the family and the region they come from.

Methods

This cross-sectional study was conducted at the King Khaled Eye Specialist Hospital (KKESH), Riyadh, Saudi Arabia. We included the parents of children ≤18 years who had a positive genetic test result for their ocular condition that indicated autosomal recessive inheritance and who received genetic counseling by a genetic counselor or a consultant in genetic ocular disease at KKESH between November 2019 and August 2020. The genetic counseling session constituted several components: collecting in-depth medical and family histories, undergoing genetic testing, disclosing the test results, evaluating the risk of the condition reoccurring, providing the couples with a preventive plan, and offering emotional and psychological support. The estimated sample size was 50 families; however, only 30 completed the interview. This study was approved by the KKESH Institutional Review Board (2101-P) and adhered to the tenets of the Declaration of Helsinki.

Questionnaire validation and design

The questionnaire validity was tested through face validity which "refers to expert opinion concerning whether the scale items measures what is intended to measure."[4] the first part of the questionnaire obtained information about the personal and demographic characteristics of participants, who were asked about the degree of consanguinity [Table 1]. The second part of the questionnaire assessed the knowledge of basic genetic information and attitude toward prevention using the following format.

Knowledge

Five multiple-choice questions with one correct answer were used in this section. These questions were used to assess:

- Basic understanding of the concept of a gene and the degree of genetic sharing within a family (2 items). The question was structured as:
  - A person shares half of their genes with their parents and siblings (agree, disagree, and I don't know)
  - The gene is a (protein, DNA, and disease).
- The parents understanding of the genetic inheritance pattern (2 items). The questions were structured as:
  - Healthy parents can have a child with an inherited disease (agree, disagree, and I don't know)
  - If the disease is transmitted in a recessive pattern it means (pathogenic mutation transmission from both parents, from the father, from the mother).
- Knowledge of the risk of recurrence in the next pregnancy (recessive inheritance pattern) (1 item). The question was structured as:
  - If you were to have another child, what are the chances of recurrence in the next pregnancy (50%, 25%, I don't know).

Table 1: Characteristics of study participants (*n*=30)

Characteristic	Count, *n* (%)
Age (years)	
<35	17 (56.7)
≥35	13 (43.3)
Gender of parent	
Female	22 (75)
Male	8 (25)
Gender of patient	
Female	17 (56.7)
Male	13 (43.3)
Education of respondent	
Uneducated	0
Elementary	1 (3.3)
Intermediate	0
Secondary	3 (10.0)
Graduate	23 (76.7)
Postgraduate	3 (10.0)
Number of siblings	
0	3 (10.0)
1	10 (33.3)
2	8 (26.7)
3	4 (13.3)
4	2 (6.7)
5	2 (6.7)
6	1 (3.3)
Parents relative degree	
Double cousin	7 (23.3)
First cousin	12 (40.0)
Second cousin	6 (20.0)
Same tribe	4 (13.3)
Not relatives	1 (3.3)

Attitude

This section comprised 5 questions with responses graded on a 3-point Likert scale (agree = 2, neutral = 1, and disagree = 0). These items assessed attitudes toward genetic counseling and prevention.

- Two questions queried parent's attitudes toward prevention in the family. It included questions about sharing their genetic information with the relatives and considering preimplantation genetic diagnosis (PGD) as an option
  - I informed/will inform my family and relatives about the DNA test result

    - I'm considering PGD.
- Two questions queried the parent's attitude toward the importance of genetic counseling in reducing the prevalence of genetic disorders and the relationship of consanguinity to hereditary diseases
    - Genetic counseling will contribute to reducing the prevalence of some genetic disorders
    - Consanguinity may lead to hereditary diseases.
- One question evaluated the feeling of social stigma.
    - I faced difficulty and embarrassment in sharing the results of the genetic test with my relatives.

Satisfaction

Parent's satisfaction with genetic counseling was assessed using a validated instrument.[5]

The instrument has measured "the two key components of patient satisfaction: technical aspects of care and interpersonal relations."[5]

Statistical analysis

Data were collected using Google Forms and analyzed using IBM SPSS statistics version 20 (IBM Corp., Armonk, NY, USA). Frequencies and proportions were calculated to describe categorical variables. Chi-squared or Fisher's exact test (2-tailed) was used to compare proportions.

Results

The study sample comprised 30 participants who met the inclusion criteria and completed the questionnaires. The mean ± SD age of respondents was 34.60±4.40 34.6 years. They were mostly fathers (73.3%) and the majority (83.3%) were in a consanguineous marriage. The participants were all from Saudi Arabia (100%), and they were distributed as follows: 33.1% from the central region, 26% from the south, 16% from the north, 10% from the East, and 10% from the West. The majority (76.7%) had completed their college education, and 70% of the parents indicated that they had 0–2 children in addition to the proband. Ninety percent of the parents had undergone one genetic counseling session, and 10% had undergone two sessions. Half of the parents surveyed had a family history of eye disorders.

Outcomes after the counseling session

Knowledge

Around 37 % parents did not understand the meaning of "the gene" and its function. Eighty percent of the parents believed that if the disease is transmitted in a recessive pattern, it means that the pathogenic mutation is transmitted from both parents. Forty-six percent of the parents gave the correct answer regarding the chances of recurrence if they were to have another child.

Attitude

Sixteen percent of the parents stated that they would not share their genetic test results with their relatives and 73.3% of parents stated that they would share their genetic test results and 10% were neutral. Sixty seven percent of the parents mentioned that they would consider using preventive measures when they consider conceiving the next child. In contrast, 23.3% of parents said that they would not take any preventative measures, and 10% were neutral. 97% of participants considered genetic counseling important and 3.3% thought it was not important. Eighty eight percent of the participants stated that consanguinity may increase the risk of hereditary diseases, and 13% were neutral. Thirty three percent of parents faced difficulty in sharing the genetic test results.

Satisfaction

Ninety percent of the parents were satisfied with the genetic counselor's ability to listen to them and the way the counselor explained the genetic information.

Testing the study objectives

Parent's knowledge and their willingness to consider using preventative measures for the next pregnancy were not significantly associated with each other ($P > 0.05$) [Table 2].

There was a statistically significant association between the parents' willingness to share the results of the genetic test with their relatives and their ability to share all of the necessary information with their genetic counselor ($P = 0.01$) [Table 3].

There was no statistically significant association between the parental attitude and the region they came from ($P > 0.05$) [Table 4].

Discussion

The outcomes of our study indicate that there was a high percentage of knowledge and a high educational level about genetic diseases in the study population. However, there was a

Table 2: Correlation between parents knowledge by the parent after genetic counseling and their willingness to consider using preventative measures in the next pregnancy

Item	*P*
Healthy parents can have a child with an inherited disease	0.519
A person shares half of their genes with their parents and siblings	>0.999
Understood the definition of the gene	>0.999
If the disease is transmitted in a recessive pattern, it means	0.630
If you were to have another child, what are the chances for recurrence?	0.687

P<0.05 is statistically significant

Table 3: Association between satisfaction with counseling and willingness to share test results with relatives

Item	*P*
I received the information I needed from my genetic counseling provider	>0.999
My provider answered all my questions	0.160
My provider listened carefully to what I said	0.545
My provider explained things in a way that was easy to understand	0.621
I was able to share all the necessary information with my provider	0.011
My provider spent enough time with me	0.712

P<0.05 is statistically significant

lack of understanding of the risk of recurrence of ocular disease in future generations. This indicates that even those who acknowledged the genetic basis did not actually understand the genetic mechanisms and the mathematical probabilities explained to them.

In an earlier study conducted by Al-Gazali, it was found that families affected by a genetic disorder often misunderstood or only partially comprehended the risk of recurrence in their future children. This research was carried out in the United Arab Emirates.[6] However, it is worth noting that the basic educational level may have evolved since then, and the population's understanding of genetic disorders could be more nuanced today.

Al-Gazali[6] also found that half of the couples understood the genetic basis of their child's condition but only 10% could recall the risk.[6] This is because they find it difficult to understand and grasp the concepts in statistical terms, such as the recurrence risk.[6] A study of retinoblastoma survivors and parents of retinoblastoma-affected children reported that these individuals might benefit from more frequent adult counseling sessions.[7]

In our study, the majority (77%) had completed their college education, and almost half of the parents indicated that they had 2 more children to none besides the proband, which is considered a small family compared to the average family size in Saudi Arabia, which was 5.9 per household in 2016.[8] This suggests that higher education levels may enhance understanding of genetic mechanisms. Furthermore, it indicates that parents of children with hereditary diseases might be aware of the risks of having more affected children. There were twelve (38%) parents who did not even know the meaning of the gene. This may affect their ability to understand certain concepts based on this information. Most parents had no problem understanding the recessive pattern of inherence, but 50% of them either overestimated the risk or did not remember the percentage. To overcome these gaps in understanding, we can use teaching aides and pamphlets to help simplifying the statistical or genetic information. In addition, follow-up appointments can ensure that the parents understand the concepts and can ask any additional questions for further clarification.

In our study, 75.34% of the sample had a positive attitude toward counseling sessions and most (97%) agreed with and were open to the role of the genetic counselor. However, there were some challenges for the parents such as the very long waiting lists and the limited availability of the genetic tests that are only performed in a few cities in the Kingdom.

Table 4: The relationship between the parental attitude and the region in Saudi Arabia they live

Item	*P*
Genetic counseling will contribute to reducing the prevalence of some genetic disorders	>0.999
I'm considering PGD next time	0.375
Consanguinity may increase the hereditary diseases	>0.999
I informed/will inform my relatives about the DNA test results	0.643
I faced difficulties and embarrassment in sharing the results of the genetic test with my relatives	0.204

P<0.05 is statistically significant. PGD: Preimplantation genetic diagnosis

An Australian study of participant expectations and satisfaction with genetic counseling found that the majority were very satisfied.[9] The outcomes of our study concur with the Australian study with 83% satisfaction rate with the genetic counseling session. In general, most of the parents answered all of the questions, listened carefully, and shared all of the necessary information with the genetic counselors. This indicates the success of the service. However, in the current study, satisfaction with counseling sessions was not correlated with their willingness to share genetic test results with their relatives ($P > 0.05$).

We found that the amount of parents' knowledge was not related to parental willingness to take preventative measures in the next pregnancy. However, we did find that the majority (66.7%) of the parents highlighted that they would take preventive measures for a following pregnancy, while 23.3% said they would not. The second group believes that this is God's will, and they do not want to interfere with it and indicated that this is the reason for their suffering rather than the abnormality in their genetic make-up. Such beliefs help alleviate feelings of guilt.[6] Some of the parents felt the burden of having one affected child and decided to stop having children until they find a treatment. One way to help parents cope with a child with a genetic disorder is by encouraging them to participate in support groups. Genetic counselors can help them accept the facts and make them comfortable communicating with others. At KKESH, there is a support group that is supervised by a consultant.

We found that 23% of our study sample did not consider preventative options for multiple reasons. Many of the parents were not willing to undergo PGD because they were afraid of switching their embryos with other different embryos during the procedure. The role of the genetic counselor could be important in such cases as they can explain the PGD procedure and that *in vitro* fertilization is done with utmost care to ensure that the embryos are not mixed. Furthermore, they can explain the steps in a Layman's terms and provide them with facts and percentages of the accuracy of the procedure. Patients also reported other challenges such as the long processes in the governmental hospitals that provide the PGD service. In some cases, parents were unaware about the PGD service at private clinics. To address this situation, we suggest having a referral system between governmental and private hospitals, which can speed up the process, although the process can also be a financial burden to the patients.

We found that 21 parents (73%) were willing to inform their relatives about the results of the genetic test. In contrast, 33.3% of the study sample was uncomfortable sharing the test results with their relatives. Some of these parents said that their relatives would not understand the situation, others said that they do not

want their child to be stigmatized by their relatives, and many parents considered this as a very sensitive and private issue. When asked about family history, some parents were isolated from their relatives. This could be because of their feeling of frustration or worrying about their children's future. Some families could share their results with the maternal relatives but not with the paternal relatives to avoid any blame or stigma. A previous study reported that the level of closeness between family members helps patients share their feelings comfortably.[10]

There are some limitations to our study including the small sample size and the short duration. These drawbacks may have affected the outcomes results. In addition, most of the participants in our study were highly educated, and we cannot definitively state whether the knowledge on genetics/inheritance they gained was from the counseling session or from another source. Future studies should assess the knowledge before counseling and after counseling and the effects of using search engines on the patient knowledge.

Conclusion

Ocular genetics is still evolving and counselors need to keep abreast of current developments. During the interviews, most of the parents asked about the latest research in regard to their diseases, and some parents who participated in one counseling session reported that there were many queries left in their minds when they left the session. Moreover, parents usually forget their questions due to the long annual waiting list, and things remain unclear to them. Hence, we recommend developing an Arabic website and printed materials for updates on genetic research. In addition, there is a need to provide more genetic counselors for follow-up sessions for patients, so they have more opportunities to have their concerns and queries addressed.

Acknowledgment

We express our appreciation to our colleagues, Dr. Kawather Karkour, Dr. Deepak Edward, Dr. Khaled Abo Amroo, Dr. Moeen AlSayed, Mr. Mohammad AlWadani, Ms. Gulsan Karbani and Ms. Reem Alkhodair who provided their expertise.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Rosenberg T. Epidemiology of hereditary ocular disorders. Dev Ophthalmol 2003;37:16-33.
2. Kotb AA, Hammouda EF, Tabbara KF. Childhood blindness at a school for the blind in Riyadh, Saudi Arabia. Ophthalmic Epidemiol 2006;13:1-5.
3. Alliance G. Genetic Alliance; The New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services. Understanding Genetics: A New York, Mid-Atlantic Guide for Patients and Health Professionals. Washington (DC): Genetic Alliance; 2009. CHAPTER 5, GENETIC COUNSELING. Available from: https://www.ncbi.nlm.nih.gov/books/NBK115552/. [Last retrieved on 2020 Oct 19].
4. Rattray J, Jones MC. Essential elements of questionnaire design and development. J Clin Nurs 2007;16:234-43.
5. Zellerino B, Milligan SA, Brooks R, Freedenberg DL, Collingridge DS, Williams MS. Development, testing, and validation of a patient satisfaction questionnaire for use in the clinical genetics setting. Am J Med Genet C Semin Med Genet 2009;151C:191-9.
6. Al-Gazali LI. Attitudes toward genetic counseling in the United Arab Emirates. Community Genet 2005;8:48-51.
7. Hill JA, Gedleh A, Lee S, Hougham KA, Dimaras H. Knowledge, experiences and attitudes concerning genetics among retinoblastoma survivors and parents. Eur J Hum Genet 2018;26:505-17.
8. General Authority for Statistics. Available form: https://www.stats.gov.sa/en/node. [Last accessed on: 2023 Aug 25].
9. Davey A, Rostant K, Harrop K, Goldblatt J, O'Leary P. Evaluating genetic counseling: Client expectations, psychological adjustment and satisfaction with service. J Genet Couns 2005;14:197-206.
10. Balobaid A, Qari A, Al-Zaidan H. Genetic counselors' scope of practice and challenges in genetic counseling services in Saudi Arabia. Int J Pediatr Adolesc Med 2016;3:1-6.