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# Knowledge of genetic eye diseases and genetic services and attitudes toward genetic testing and gene therapy

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## Abstract:

**PURPOSE:** Genetic eye diseases are among the top ten causes of ocular health burden. Asia accounts for nearly two-thirds of the global burden of genetic eye diseases. A great deal of resources is being invested in genetic research and development of genetic services including gene testing laboratories and genetic counseling in India. These efforts will be meaningful only if the public and clinicians are aware of their existence. This study aimed to understand the level of knowledge about genetic eye diseases and genetic services and attitudes toward genetic testing and gene therapy in four groups of participants (undergraduate medical students, paramedical staff, non-ophthalmologist doctors, and the general public).

**MATERIALS AND METHODS:** This was a cross-sectional survey in India. Four hundred questionnaires were analyzed from the four groups of participants. Knowledge score was calculated for the different questions. To bring out the differences across the groups, Chi-square test was done with a post hoc Mann–Whitney U-test and Kruskal–Wallis test.  $P < 0.05$  was taken as statistically significant.

**RESULTS:** The level of awareness about genetic eye diseases was better among undergraduate students, doctors, and paramedical staff compared to the general public ( $P < 0.001$ ). The majority across all three groups had a positive attitude toward genetic testing and gene therapy. However, most of the participants across all groups were not aware of the genetic facilities available in our country.

**CONCLUSION:** This study shows a positive attitude toward genetic medicine. However, there is a need to improve public awareness about genetic eye diseases and facilities available for genetic testing and gene therapy.

## Keywords:

Attitudes, genetic eye diseases, genetic testing, gene therapy, genetic services, knowledge

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## Introduction

The World Health Organization has identified genetic eye diseases as one of the top ten causes of ocular health burden.<sup>[1]</sup> With the changing demographics, noncommunicable diseases are on the rise. Genetic diseases constitute a significant portion of such noncommunicable diseases.<sup>[2]</sup> India is a country where a large number of communities still favor

consanguineous marriages, which increases the risk of genetic disease transmission.<sup>[3,4]</sup> It has been reported that nearly 30% of the patients with genetic eye diseases have a family history of consanguinity in India.<sup>[4]</sup> The prevalence of retinitis pigmentosa (RP), a very common genetic eye disease, is 1 in 930 in urban and 1 in 372 in rural South Indian population aged 40 years and over - a number significantly higher than in the Western population.<sup>[5]</sup> However, genetic eye diseases continue to receive very little attention from health planners, doctors, and

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the general public due to the misconception that they are rare diseases.

A great deal of resources is being invested worldwide and in our country in ocular genetics.<sup>[6]</sup> There has been tremendous growth in the knowledge about genetic eye diseases in recent years. There are many gene therapy trials going on in genetic eye diseases such as Leber's congenital amaurosis, choroideremia, Usher syndrome, and X-linked retinoschisis.<sup>[7-9]</sup> Genetic testing has become more widespread than it was before. Genetic testing helps in accurately diagnosing the gene at fault, predicting the risk of transmission to the offsprings, and guiding genetic counseling and gene therapy.<sup>[10]</sup> Genetic counselors educate individuals and families about the underlying biology of the disease and its inheritance patterns, and help them cope with the medical and psychological implications.<sup>[11]</sup> Gene therapy is a novel drug delivery method where viral or nonviral vectors are used to deliver the target gene into the patient's cells.<sup>[12]</sup> A breakthrough in the field of ocular genetics was achieved when the Food and Drug Administration approved gene therapy for patients with RPE65-related retinal degeneration.<sup>[13]</sup> However, these discoveries will be meaningful only if the target population is aware of these diseases and the available options for testing and treatment.

This study aims to understand the level of knowledge about genetic eye diseases and genetic services and attitudes toward genetic testing and gene therapy of undergraduate medical students, paramedical staff, non-ophthalmologist doctors, and the general public.

## Materials and Methods

The study was conducted between July 2020 and January 2021. This study was conducted according to the Declaration of Helsinki and the National Ethical Guidelines for Biomedical and Health Research involving Human Participant laid down by the Indian Council of Medical Research. Approval from the Ethics Committee of All India Institute of Medical Sciences (AIIMS) (AIIMS/MG/IEC/2020-21/26), Mangalagiri, was obtained to carry out this research.

### Study population

Four groups of volunteers were chosen: undergraduate medical students, paramedical staff (optometrists, nurses, and laboratory technicians), non-ophthalmologist doctors, and the general public.

### Sample size calculation

Sample size was calculated using the formula

$$(n) = \frac{(Z_{1-\alpha/2})^2 \times (p) \times (q)}{(d)^2}, \text{ where } Z_{1-\alpha/2} = 1.96$$

(at 95% confidence interval or 5% level of significance),  $p = 0.35$  (35% prevalence assumed),  $q = 1 - p = 0.65$ , and  $d = 0.05$  (5% margin of error or precision). Using this, the sample size was calculated to be 350. Considering a 10% dropout of the study participants, the sample size was calculated to be 385.

### Study design

This was a cross-sectional study conducted in India. The questionnaire consisted of three parts with multiple-choice and Likert scale-based questions. Some open-ended questions were also included. Part A of the questionnaire had 11 questions to capture the demographic details of the participants. Part B (17 questions) aimed to test the level of knowledge of the participants about genetics in general and genetic eye diseases, in particular genetic testing and gene therapy. Part C (14 questions) was designed to assess the attitudes toward genetic testing and gene therapy.

These self-administered questionnaires were distributed randomly to the participants using an online link or on paper to those who do not have access to the internet. Informed consent was taken digitally or on paper, and each participant was not allowed to take the survey more than once. The data were entered and organized in a spreadsheet and exported to SPSS version 23.0 software (SPSS Inc., IBM, Chicago, IL, USA).

### Statistics

Descriptive statistics (demographic details) were presented as counts and percentages. For assessment of the knowledge, each correct response was scored as 1 and an aggregated knowledge score was calculated. This was summarized as the median and interquartile range. To bring out the differences in the knowledge score across the groups, a Chi-square test was carried out with a *post hoc* Mann-Whitney *U*-test and Kruskal-Wallis test. For Likert scale-based questions, scores 1 and 2 were taken for disagreement and 4 and 5 for agreement with the question. A score of 3 was taken as no opinion. SPSS 23.0 software (SPSS Inc., IBM, Chicago, IL, USA) was used for statistical analysis.  $P < 0.05$  was taken as statistically significant.

## Results

A total of 400 questionnaires were analyzed (71 undergraduate students, 77 paramedical staff, 105 nonophthalmology doctors, and 147 members of the general public). Two hundred and eighty questionnaires were answered online and 120 on paper. The overall response rate was 43.7%.

## Demography

The demographic profile of the participants is shown in Table 1. Sixty-two percent of the participants were from cities, 20% from towns, and 18% from the villages of the country. More than 90% of the participants in each group had at least an undergraduate degree. Only 66.7% of the general public had a background of learning genetics at school/college. The majority of the undergraduate students, paramedical staff, and doctors got their information about genetics through formal teaching at school/college. However, the members of the general public were more diverse in this respect (48.3% from school/college teaching, 17.7% from newspapers, 20.4% from social media, and 13.6% from hearsay information).

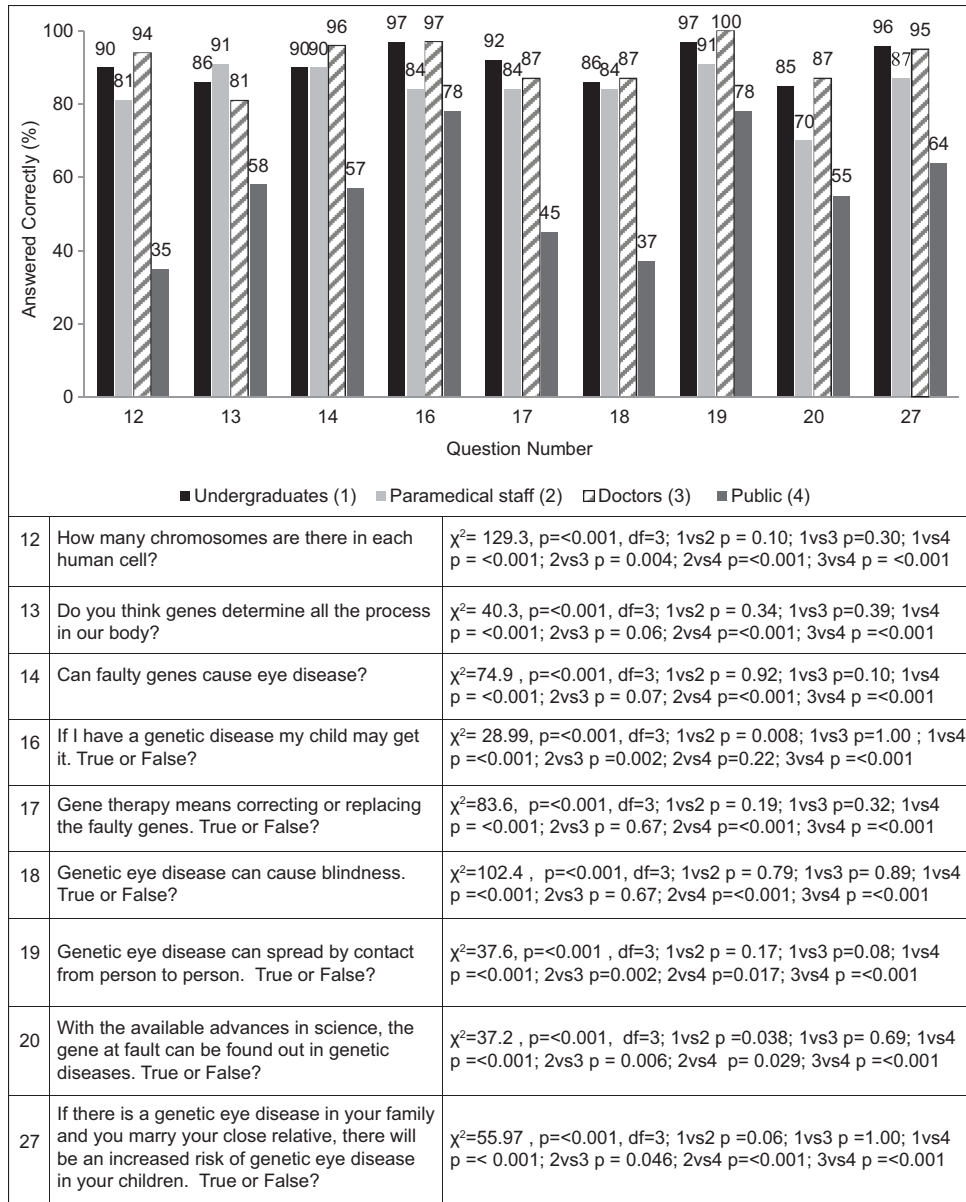
## Knowledge about genetics, genetic eye diseases, genetic testing, and gene therapy

In general, the level of understanding about basic concepts in genetics was better in the first three groups compared to the members of the general public [ $P < 0.001$ ; Figure 1]. However, it is surprising to note that nearly 8% of the undergraduate students, 13% of the doctors, and 16% of the paramedical staff were not sure what gene therapy involves. Ten percent of the undergraduates and paramedical staff did not even know that faulty genes can cause eye diseases. The most commonly cited examples of genetic eye diseases were retinoblastoma, myopia, congenital cataract, ectopia lentis, color blindness, etc., Only nine participants were aware of a genetic eye disease which gene therapy has been tried for.

**Table 1: Demographic profile of the participants**

	Undergraduate students, n (%)	Paramedical staff, n (%)	Doctors, n (%)	Public, n (%)
Age, mean±SD (years)	20±1.7	28.6±3.8	33±4.7	37.4±11.8
Gender				
Male	34 (47.9)	33 (42.9)	61 (58.1)	94 (63.9)
Female	37 (52.1)	44 (57.1)	44 (41.9)	53 (36.1)
Education				
Primary schooling	0	0	0	1 (0.7)
Secondary schooling	3 (4.2)	2 (2.6)	0	8 (5.4)
Undergraduation	65 (91.6)	47 (61.0)	6 (5.7)	67 (45.6)
Postgraduation	3 (4.2)	28 (36.4)	99 (94.3)	71 (48.3)
Religion				
Christian	7 (9.9)	14 (18.2)	8 (7.6)	10 (6.8)
Hindu	54 (76.1)	57 (74)	92 (87.6)	125 (85)
Muslim	9 (12.7)	2 (2.6)	1 (0.9)	7 (4.8)
Atheist	1 (1.3)	2 (2.6)	2 (1.9)	3 (2.0)
Others	0	2 (2.6)	2 (1.9)	2 (1.4)
Annual income (lakhs)				
Nil	66 (92.9)	10 (12.9)	4 (3.8)	20 (13.6)
0-5	4 (5.6)	29 (37.7)	11 (10.5)	51 (34.7)
5-10	0	33 (42.9)	25 (23.8)	34 (23.1)
>10	1 (1.4)	5 (6.5)	65 (61.9)	42 (28.6)
Taught genetics at school/college				
Yes	68 (95.8)	73 (94.8)	101 (96.2)	98 (66.7)
No	3 (4.2)	4 (5.2)	4 (3.8)	49 (33.3)
If so up to what level?				
Primary schooling	0	0	0	14 (14.3)
Secondary schooling	14 (20.6)	7 (9.6)	6 (5.9)	46 (46.9)
Undergraduation	54 (79.4)	52 (71.2)	21 (20.8)	28 (28.6)
Postgraduation	0	14 (19.2)	74 (73.3)	10 (10.2)
Information about genetics is from?				
School/college education	67 (94.4)	72 (93.5)	102 (97.1)	71 (48.3)
Newspaper/magazines	1 (1.4)	1 (1.3)	2 (1.9)	26 (17.7)
Social media	2 (2.8)	3 (3.9)	1 (0.9)	30 (20.4)
Hearsay	1 (1.4)	1 (1.3)	0	20 (13.6)
Any family member with genetic disorder?				
Yes	9 (12.7)	8 (10.4)	9 (8.6)	21 (14.3)
No	62 (87.3)	69 (89.6)	96 (91.4)	126 (85.7)
Use Internet for medical information				
Yes	67 (94.4)	76 (98.7)	98 (93.3)	119 (80.9)
No	4 (5.6)	1 (1.3)	7 (6.7)	28 (19.0)

SD=Standard deviation



**Figure 1:** Participant knowledge about basic concepts in genetics and genetic eye diseases. This figure shows that the level of understanding about basic concepts in genetics was better in the first three groups compared to the members of the general public.

Table 2 shows the distribution of knowledge scores based on gender, occupation, education, presence of genetic disease in the family, genetic teaching in school/college, and use of the internet for medical information.

The knowledge about the genetic services available in the country was uniformly poor across the groups [Figure 2]. While the majority said they would see an eye doctor, a small proportion was unsure who to consult in case of genetic eye disease.

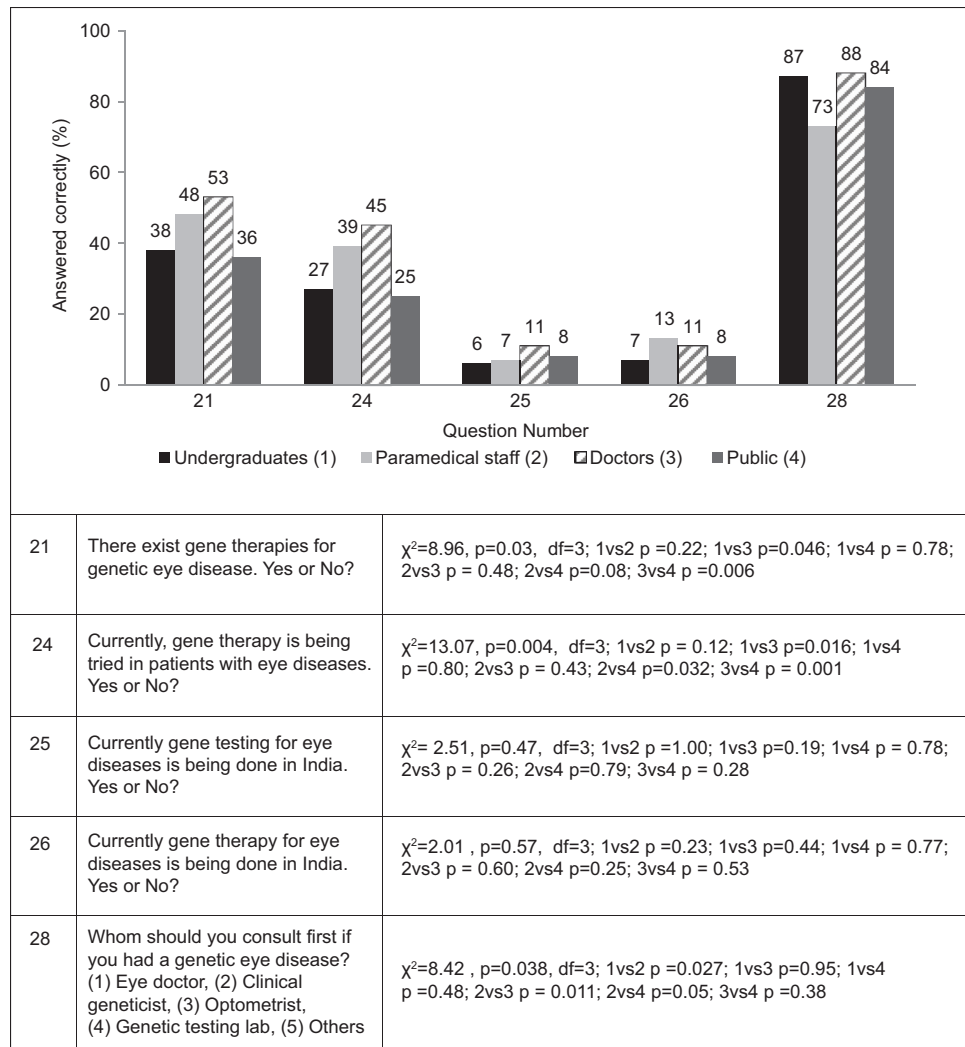
### Attitudes toward genetic testing and gene therapy

There was a positive attitude toward genetic testing and gene therapy across all groups of participants. The

majority of the participants neither felt that genetic testing is a waste of resources if treatment is not available nor did they feel gene therapy was unethical. The majority of the undergraduates and doctors were ready to undergo genetic testing and gene therapy if they had a genetic eye disease. However, only 63% of the general public were ready to undergo gene testing and a much lesser proportion gene therapy (49%) if they had a genetic eye disease [Figure 3].

### Discussion

The current study shows that the level of understanding about basic concepts of genetics, genetic eye diseases, genetic testing, and gene therapy was better in the first



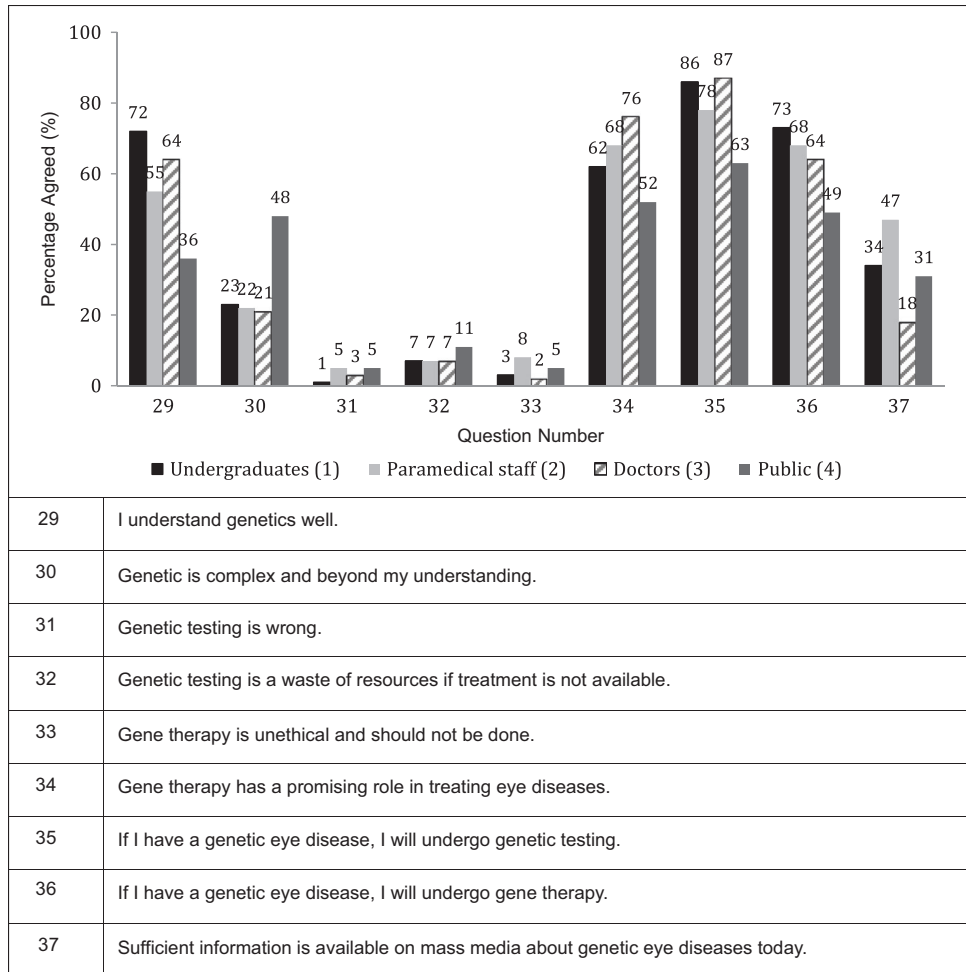
**Figure 2:** Participant awareness about the genetic services available. In general, the majority of the participants across all the groups were not aware of the facilities available.

three groups (undergraduate students, paramedical staff, and doctors) compared to the members of the general public. However, the awareness about the testing facilities available in the country was poor across all the groups. In general, there was a positive attitude toward genetic testing and gene therapy among all groups of participants.

We have seen extensive development in the field of ocular genetics in the past decade worldwide. India has a large population of patients with Mendelian genetic eye diseases such as RP, Leber's congenital amaurosis, and Stargardt's disease. Due to the widespread practice of consanguineous marriages, this number is rising year after year.<sup>[4,5]</sup> Most of these cases are concentrated in the villages and backward areas where there is a general lack of knowledge about genetic diseases and lack of facilities for testing for the same.

In the current study, we have seen that there was a general lack of knowledge about basic concepts in genetics

among the members of the general public. Undergraduate students, paramedical staff, and doctors showed a good understanding of these basic concepts possibly owing to their association with the medical profession. However, even these three groups of participants did not display the expected level of knowledge. Nearly 8% of the undergraduate students, 13% of the doctors, and 16% of the paramedical staff were not sure what gene therapy involves and 10% of the undergraduates and paramedical staff did not even know that faulty genes can cause eye diseases. Most of the participants in these three groups could only name myopia and retinoblastoma as examples of genetic eye diseases, while these are not even the classical examples of Mendelian genetic eye diseases. Although basic concepts about genetics are included in the curriculum in primary and secondary schooling, the members of the general public could not answer basic questions about genes and the role they play. The following could be the reasons for this teaching-learning gap in our country as stated by Chattopadhyay: learning biology



**Figure 3:** Participant attitudes to genetic testing and gene therapy. Most of the participants had a positive attitude toward genetic testing and gene therapy.

involves only memorization of facts at both the school and college levels, teachers tend to avoid these topics to the students because they are considered complex, and examinations permit students to answer alternative questions.<sup>[14]</sup> Lewis *et al.* and Marbach-Ad have reported similar gaps in the Western teaching methodology as well.<sup>[15,16]</sup> Instructional courses based on the principles of active learning and the use of multimedia in teaching can be more effective in overcoming these hurdles.<sup>[17,18]</sup> Helping students understand genetics conceptually and not as mere facts is necessary to bridge this gap.

The internet has emerged as a powerful tool for disseminating information. We observed a significant difference in the knowledge scores between people who used the internet for medical information versus those who did not ( $P = 0.003$ ). Most of the participants felt genetic eye diseases were not well publicized. The credibility of the information that is available on the internet has always been questioned by the public.<sup>[19]</sup> Hence, creating websites wherein authentic information (about genetic eye diseases and facilities available for testing, treatment, and counseling) is

updated from time to time may be an effective and easy way of disseminating knowledge.

Research has reached the stage where we are now exploring gene therapy for genetic eye diseases.<sup>[7-9]</sup> Recently, the FDA approved gene therapy for patients with RPE65-related retinal degeneration, but people seem to be largely unaware of these developments.<sup>[13]</sup> Genetic laboratories equipped with next-generation sequencing, targeted panel testing, and whole-exome sequencing are available in India today.<sup>[6]</sup> However, the majority of the participants across all groups were not aware of this. This knowledge gap is not because the participants were against genetic testing or gene therapy as it can be seen that the majority across all groups were ready to undergo genetic testing/therapy if they had a genetic eye disease. This reflects the sheer lack of information about these services in the country even among the clinicians. This ignorance has resulted in the underutilization of these services. Other factors that contribute to this are inequality in the distribution of genetic services, cost of testing, and lack of formal referral pathways.<sup>[20,21]</sup>

**Table 2: Distribution of knowledge score**

	Median	IQR	P
Gender			
Male	11.00	9-13	0.007
Female	12.00	10-13	
Occupation			
Undergraduates	12.00	11-13	<0.001
Paramedical staff	12.00	10-13	
Doctors	13.00	12-14	
Public	9.00	7-11	
Education			
Secondary schooling	9.00	6-11	0.002
Undergraduation	11.00	9-13	
Postgraduation	12.00	10-14	
Annual income (lakhs)			
Nil	12.00	10-13	0.002
0-5	10.00	8-12	
5-10	11.00	10-13	
>10	12.00	10-14	
Were you taught genetics at school/college?			
Yes	12.00	10-13	<0.001
No	9.00	7-11	
Does any family member have genetic disease?			
Yes	11.00	9-13	0.611
No	11.00	9-13	
Do you use internet for medical information?			
Yes	11.50	10-13	0.003
No	9.50	8-11.75	

IQR=Interquartile range

Previous studies from other countries have shown a general lack of knowledge about genetic testing among the general public in developing and developed countries. According to the 2013 U.S. Health Information National Trends Survey, only 35.6% of the participants were aware of the direct-to-consumer genetic testing.<sup>[22]</sup> A systematic review published by Hann *et al.*<sup>[23]</sup> revealed low awareness and knowledge of genetic counseling/testing among minority groups including African Americans, Asian Americans, and Hispanics. In a large study conducted on health-care workers, Baars *et al.* found a gross deficiency in the knowledge levels of genetics in many non-geneticist health-care providers.<sup>[24]</sup> A similar study conducted in the UK showed that optometry students and primary eye care professionals were better informed about genetic eye diseases than the general public. However, all groups of participants showed a lack of knowledge about the genetic services available in the UK.<sup>[25]</sup> This shows that these deficiencies are a global problem.

The limitations of this study are those inherent to questionnaire studies: (i) recall bias, (ii) the participants could have given desirable answers rather than the true

answers, and (iii) attitudinal bias where more positive people are more likely to take the survey.

The results of this study call for a more concerted effort in disseminating information about genetic eye diseases, genetic testing, and gene therapy among clinicians, paramedical staff, students, and the general public. In addition, there is a need to create formal referral pathways for referring patients with genetic eye diseases like in the West.<sup>[26]</sup> Genetic medicine in India can make meaningful contributions to health care only when the public and physicians are well informed about the potential of this science.

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### Conflicts of interest

The authors declare that there are no conflicts of interests of this paper.

### References

- Mohan S, Satagopan U, Nagasamy S, Sundaram N. Genetics and susceptibility of retinal eye diseases in India. In Prakash G & Iwata T (Eds.). *Advances in Vision Research, Volume II, Essentials in Ophthalmology*. Singapore: Springer; 2019. p. 147-68.
- Verma IC. Burden of genetic disorders in India. *Indian J Pediatr* 2000;67:893-8.
- Nirmalan PK, Krishnaiah S, Nutheti R, Shamanna BR, Rao GN, Thomas R. Consanguinity and eye diseases with a potential genetic etiology. Data from a prevalence study in Andhra Pradesh, India. *Ophthalmic Epidemiol* 2006;13:7-13.
- Kumaramanickavel G, Joseph B, Vidhya A, Arokiasamy T, Shridhara Shetty N. Consanguinity and ocular genetic diseases in South India: Analysis of a five-year study. *Community Genet* 2002;5:182-5.
- Sen P, Bhargava A, George R, Ve Ramesh S, Hemamalini A, Prema R, *et al.* Prevalence of retinitis pigmentosa in South Indian population aged above 40 years. *Ophthalmic Epidemiol* 2008;15:279-81.
- Aggarwal S, Phadke SR. Medical genetics and genomic medicine in India: Current status and opportunities ahead. *Mol Genet Genomic Med* 2015;3:160-71.
- Simonelli F, Maguire AM, Testa F, Pierce EA, Mingozzi F, Bennicelli JL, *et al.* Gene therapy for Leber's congenital amaurosis is safe and effective through 1.5 years after vector administration. *Mol Ther* 2010;18:643-50.
- Goldmann T, Overlack N, Möller F, Belakhov V, van Wyk M, Baasov T, *et al.* A comparative evaluation of NB30, NB54 and PTC124 in translational read-through efficacy for treatment of an USH1C nonsense mutation. *EMBO Mol Med* 2012;4:1186-99.
- MacLaren RE, Groppe M, Barnard AR, Cottrill CL, Tolmachova T, Seymour L, *et al.* Retinal gene therapy in patients with choroideremia: Initial findings from a phase ½ clinical trial. *Lancet* 2014;383:1129-37.
- Burke W, Zimmern RL, Kroese M. Defining purpose: A key step in genetic test evaluation. *Genet Med* 2007;9:675-81.
- Sutherland JE, Day MA. Genetic counseling and genetic testing in ophthalmology. *Curr Opin Ophthalmol* 2009;20:343-50.
- Singh VK, Tripathi P. Gene therapy in ocular diseases. *Indian J Ophthalmol* 2002;50:173-81.

13. Russell S, Bennett J, Wellman JA, Chung DC, Yu ZF, Tillman A, *et al.* Efficacy and safety of voretigene neparvovec (AAV2-hRPE65v2) in patients with RPE65-mediated inherited retinal dystrophy: A randomised, controlled, open-label, phase 3 trial. *Lancet* 2017;390:849-60.
14. Chattopadhyay A. Understanding of genetic information in higher secondary students in northeast India and the implications for genetics education. *Cell Biol Educ* 2005;4:97-104.
15. Lewis J, Leach J, Wood-Robinson C. Chromosomes: The missing link-young people's understanding of mitosis, meiosis, and fertilization. *J Biol Educ* 2000;34:189-99.
16. Marbach-Ad G. Attempting to break the code in students' comprehension of genetic concepts. *J Biol Educ* 2001;35:183-9.
17. Smith MK, Wood WB. Teaching genetics: Past, present, and future. *Genetics* 2016;204:5-10.
18. Starbek P, Starčić Erjavec M, Peklaj C. Teaching genetics with multimedia results in better acquisition of knowledge and improvement in comprehension. *J Comput Assist Learn* 2010;26:214-24.
19. Bernhardt JM, Lariscy RA, Parrott RL, Silk KJ, Felter EM. Perceived barriers to Internet-based health communication on human genetics. *J Health Commun* 2002;7:325-40.
20. Balarajan Y, Selvaraj S, Subramanian SV. Health care and equity in India. *Lancet* 2011;377:505-15.
21. Engel N, Ganesh G, Patil M, Yellappa V, Pant Pai N, Vadnais C, *et al.* Barriers to point-of-care testing in India: Results from qualitative research across different settings, users and major diseases. *PLoS One* 2015;10:e0135112.
22. Agurs-Collins T, Ferrer R, Ottenbacher A, Waters EA, O'Connell ME, Hamilton JG. Public awareness of direct-to-consumer genetic tests: Findings from the 2013 U.S. Health Information National Trends Survey. *J Cancer Educ* 2015;30:799-807.
23. Hann KE, Freeman M, Fraser L, Waller J, Sanderson SC, Rahman B, *et al.* Awareness, knowledge, perceptions, and attitudes towards genetic testing for cancer risk among ethnic minority groups: A systematic review. *BMC Public Health* 2017;17:503.
24. Baars MJ, Henneman L, Ten Kate LP. Deficiency of knowledge of genetics and genetic tests among general practitioners, gynecologists, and pediatricians: A global problem. *Genet Med* 2005;7:605-10.
25. Ganne P, Garrioch R, Votruba M. Perceptions and understanding of genetics and genetic eye disease and attitudes to genetic testing and gene therapy in a primary eye care setting. *Ophthalmic Genet* 2015;36:50-7.
26. Moore T, Burton H. Genetic Ophthalmology in Focus: Report of a UKGTN Working Party. Foundation for Genomics and Population Health; 2008.