Original Article

Assessment of knowledge, awareness, and attitude toward genetic diseases among medical students studying in a tertiary health-care teaching hospital in Uttarakhand, India

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ABSTRACT

There is a high prevalence of genetic disorders in India. Factors that favor high prevalence are the large population, high birth rate, and consanguineous marriage in many communities. The majority of physicians rely upon knowledge of medical schools gained at the undergraduate level. Currently, at the undergraduate level, medical genetics is being taught by different departments in different semesters. It creates a lack of continuity and integrity. This study identified that medical students have limited knowledge about basic medical genetics, moderate awareness regarding the epidemiological and social impact of genetic diseases, and an optimistic attitude toward the management of genetic diseases in society. Curricular teaching and practical exposure might help increase knowledge and awareness and inculcate practicality in students' attitudes.

Key words: Attitude, Awareness, Genetic disorders, Knowledge, Students

There is a high prevalence of genetic disorders in India; factors that favor high prevalence are the large population, high birth rate, and consanguineous marriage in many communities [1]. The prevalence of multifactorial chronic diseases such as coronary artery disease and hypertension is also high [1]. In a study conducted in AIIMS, New Delhi, it was found that more deaths were caused by sepsis and few by congenital disorders in the perinatal period in the 1970s, and the pattern was reversed by the late 1990s [2]. Genetic disorders constitute the second most common cause of infant mortality in India with a prevalence of 25–60/1000 births [3].

There is no national public health program for the screening of genetic disorders in newborns [4]. Prevention is the only practical option for most genetic diseases in developing countries like India. Genetic diseases can be prevented by community education, population screening, pre-marital genetic counseling, and pre-natal diagnosis [5,6]. These facilities should be available in the primary health-care center, and our primary health-care physicians should have adequate knowledge and training in medical genetics. The WHO also emphasizes genetic services in primary health care [7].

Access this article online	
Received- 26 May 2023 Initial Review- 03 Jun 2023 Accepted- 06 Jun 2023	Quick Response code
DOI: 10.32677/ijch.v10i7.4066	

The majority of physicians excluding post-graduation programs rely upon knowledge of medical schools gained at the undergraduate level [8]. A study conducted by SGPGI, Lucknow, found that knowledge about medical genetics was inadequate among clinicians [9]. Currently, at the undergraduate level, medical genetics is being taught by different departments in different semesters. It creates a lack of continuity and integrity of knowledge about genetics.

This study will show the present scenario of education in medical genetics in medical colleges in India. Reduction in the burden of genetic diseases can only be achieved if we give adequate training to medical undergraduate students about medical genetics. Hence, graduate medical students will have skills for the identification of genetic disorders in the community. There is a need for a separate medical genetics department in medical colleges, and the curriculum should be framed in such a way that physicians of any specialty can identify common genetic disorders and refer the patient to genetic centers [9]. The objective of this study was to assess the knowledge of medical students regarding medical genetics, their awareness regarding its epidemiological and social impact, and their attitude of medical students toward medical genetics among the medical students.

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METHODOLOGY

This was a cross-sectional study conducted in a tertiary care teaching hospital in Uttarakhand. Through convenient sampling, undergraduate medical students (professionals 1 through 4) and interns studying at a tertiary health-care teaching hospital in Uttarakhand were invited to participate in the study. Participants were enrolled in the study with due well-informed e-consent detailing the need and implications of the study collected through a Google Forms. This study was conducted with due ethical approval from the Institutional Research Review Board and Institutional Ethics Committee (Reg. No.: EC/NEW/Inst/2020/1046) received through letter number - AIIMS/IES/21/588 dated – November 20, 2021. This study was carried out by all relevant guidelines and investigations.

A structured Google Forms was used to survey the sociodemographic details of the participants and an investigator-designed validated questionnaire to assess the participants:

a. Knowledge of medical genetics: Knowledge of medical students about the essentials of medical genetics was assessed through six objective questions (item numbers 3, 12, 13, 14, 15, and 16) about the basics of genetic diseases and their clinical outcomes
b. Awareness regarding the epidemiological and social impact of genetic diseases: Awareness of students regarding the epidemiological and social impact of genetic diseases on potential strategies to enhance societal awareness regarding the same was assessed by five forward-directed (item numbers 1, 2, 7, 9, and 11) and one backward-directed (item number 4) Likert scale questions where higher scores were indicative of stronger agreement to the statements regarding the aforementioned
c. Attitude: The attitude of medical students toward the occurrence and management of genetic disorders in society was evaluated through two forward-directed (item numbers 5 and 10) and two backward-directed (item numbers 6 and 8) Likert scale questions where higher scores were indicative of stronger agreement to the statements regarding a health-care professional's attitude toward medical genetics.

The questionnaire was validated by calculating the scale-level CVI for the form based on its assessment by a panel of four content experts. Besides achieving relevant ratings, the items had high content validity as well which indicated that data obtained from the form adequately mapped the individual domains and were truly representative of them. The 16-item questionnaire was designed to assess knowledge, attitude, and awareness about medical genetics. Students who consent to participate were distributed the questionnaire regarding knowledge, awareness, and attitude toward medical genetics.

Outcome Measures

The outcomes of the study were as follows -

1. Knowledge of medical students regarding medical genetics reported in discrete interval variables ranging from 0 to 6
2. Awareness of medical students regarding the epidemiological and social impact of genetic diseases reported in discrete interval variables ranging from 6 to 30 with a higher score suggestive of a higher awareness about the epidemiological and social impact of genetic diseases
3. The attitude of medical students toward the occurrence and management of genetic disorders in society was reported in discrete interval variables ranging from 4 to 20 with a higher score suggestive of a more practical attitude of students toward genetic disorders in the society.

Statistical Plan Analysis

The results of the study have been reported descriptively.

RESULTS

The study involved a total of 68 students, and their distribution across different academic years is presented in Fig. 1.

The distribution of the study variables was checked using the Shapiro–Wilk test. All three variables, namely knowledge, awareness, and attitude, were found to follow a non-parametric distribution and hence non-parametric measures of central tendency and dispersion have been used to interpret the collected data. The cumulative and question-wise responses of the participants to the questionnaire are elaborated in Tables 1-3.

Table 1 provides descriptive statistics for the variables of knowledge, awareness, and attitude among the participants in the study, offering valuable insights into the distribution and central tendency of the data.

Regarding the "Knowledge" variable, the maximum score of 6 indicates the highest level of knowledge observed. The average score of 4.764 represents the typical level of knowledge among the participants. The standard deviation (SD) of 0.948 indicates the variability of scores around the mean. The 25th percentile of 4 suggests that 25% of participants scored below this value. The median score of 5 indicates that half of the participants scored at or below this value. The 75th percentile of 5 indicates that 75% of participants scored at or below this value. The interquartile range (IQR) of 1 represents the range between the 25th and 75th percentiles.

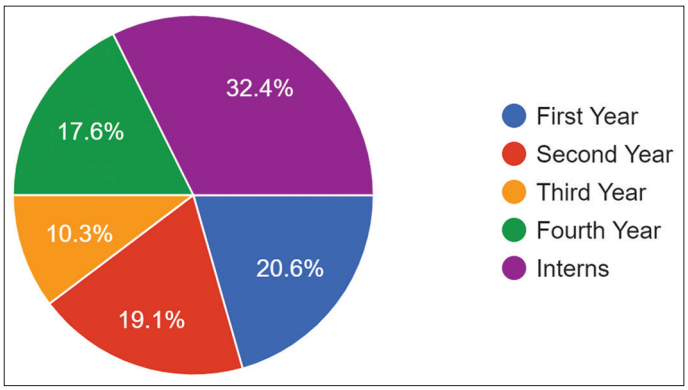

Figure 1: Year-wise distribution of students who participated in the study

Table 1: Measures of central tendency and dispersion of the knowledge, awareness, and attitude of medical students about medical genetics

Variables	Max	Mean	SD	25th Percentile	Median	75th Percentile	IQR
Knowledge	6	4.764	0.9481	4	5	5	1
Awareness	30	21.176	5.410	19.75	21	25	5.25
Attitude	20	12.338	3.290	9.75	13	15	5.25

IQR: Interquartile range

Table 2: Measures of central tendency and dispersion of each question assessing awareness of medical students regarding the epidemiological and social impact of genetic diseases

Questions	Mean	SD	25th Percentile	Median	75th Percentile	IQR
Genetic disorders are a major cause of infant mortality.	3.632	1.105	3.75	4	4	0.25
Genetic diseases can affect any person.	3.838	0.891	4	4	4	0
Numerous genetic diseases have curative therapy.	2.867	0.960	2	3	4	2
Social stigma is a factor when someone hides having a genetic disease.	4.073	0.697	4	4	4	0
Basic medical genetics knowledge should be made compulsory in primary and secondary education to enhance social awareness.	4.279	0.642	4	4	5	1
Parents are to be held accountable for getting a child with a genetic disease without a family history.	2.485	1.112	2	2	4	2

IQR: Interquartile range

Table 3: Measures of central tendency and dispersion of each question assessing the attitude of medical students toward the occurrence and management of genetic disorders in society

Questions	Mean	SD	25th percentile	Median	75th percentile	IQR
Prevention is the best management for genetic disorders in our society.	3.382	1.133	2.75	4	4	1.25
Common genetic disorders can be eradicated from society.	2.941	0.944	2	3	4	2
There is a lack of social awareness for genetic disorders.	1.691	0.579	1	2	2	1
Post-marriage and pre-conception counseling provide a better way to prevent common genetic diseases in lower socioeconomic families.	4.323	0.633	4	4	5	1

IQR: Interquartile range

For the "Awareness" variable, the maximum score of 30 reflects the highest level of awareness observed. The average score of 21.176 represents the typical level of awareness among the participants. The SD of 5.410 indicates a relatively large dispersion of scores around the mean. The 25th percentile of 19.75 suggests that 25% of participants scored below this value. The median score of 21 indicates that half of the participants scored at or below this value. The 75th percentile of 25 suggests that 75% of participants scored at or below this value. The IQR of 5.25 represents the range between the 25th and 75th percentiles.

Regarding the "Attitude" variable, the maximum score of 20 represents the highest level of attitude observed. The average score of 12.33 indicates the typical attitude level among the participants. The SD of 3.291 shows the dispersion of scores around the mean. The 25th percentile of 9.75 suggests that 25% of participants scored below this value. The median score of 13 indicates that half of the participants scored at or below this value. The 75th percentile of 15 suggests that 75% of participants scored at or below this value. The IQR of 5.25 represents the range between the 25th and 75th percentiles.

Table 2 presents the descriptive statistics for each question assessing the awareness of medical students regarding various aspects of genetic disorders in society. These statistics provide valuable information about the distribution and central tendency of the participants' responses, allowing for a comprehensive understanding of their attitudes.

For instance, consider the question "Genetic disorders are a major cause of infant mortality." The mean response reflects the average level of agreement or disagreement among the participants regarding this statement. The SD indicates the variability in their responses, suggesting the extent of consensus or diversity among the participants. The percentile values offer insights into the distribution of responses, highlighting the proportion of participants with scores below certain thresholds. The IQR provides an understanding of the spread of responses within the middle 50% of the data.

Likewise, for each question, the table 2 provides the mean, SD, percentile values, and IQR, enabling a comprehensive analysis of the participants' attitudes toward genetic disorders about the specific statements.

Table 3 presents descriptive statistics for each question that assesses the attitude of medical students regarding the occurrence and management of genetic disorders. These statistics provide valuable insights into the participants' responses, offering a comprehensive understanding of their awareness levels.

For instance, consider the question "Prevention is the best management for genetic disorders in our society." The mean response of 3.382 indicates the average level of agreement or disagreement among the participants. The SD of 1.133 suggests

a moderate amount of variability in their responses, indicating a range of opinions. The 25th percentile value of 2.75 indicates that 25% of participants responded with a score below this value, whereas the median response of 4 represents the middle value in the ordered responses. The 75th percentile of 4 suggests that 75% of participants scored at or below this value. The IQR of 1.25 reflects a moderate spread of responses within the middle 50% of the data.

Similarly, the table 3 provides the mean, SD, percentile values, and IQR for each of the other questions. These statistics provide valuable insights into the participants' awareness levels regarding genetic disorders with the specific statements mentioned in the table 3.

DISCUSSION

This study was conducted at a tertiary care teaching hospital in Uttarakhand. The total strength of the medical school affiliated with the same is 500. Of 500 medical students studying in the institute, only a total of *n*=68 responded to the survey equating to a response rate of 13.6%. Response rates to all survey methods have decreased over the past decade, but web surveys have been reported to have experienced a greater attenuation in their response rates [10-13]. The increase in research activity during COVID-19 accompanied by the restrictions on in-person data collection to ensure social distancing resulted in a surge of web survey-based research, resulting in "Survey Fatigue" among potential participants of research studies [14]. While a response rate of 40% has been reported to indicate a good survey study with precise and reliable survey results, the current study could not match the same probably owing to post-COVID survey fatigue [15].

The knowledge of medical students about basic medical genetics was less than the maximum. While the mean of the data was expected to be lesser, owing to about 40% of respondents (1st- and 2nd-year medical students) not being exposed to medical genetics and genetic diseases, the lower median does raise concerns that senior and graduating medical students also have limited knowledge of medical genetics, less than the desirable. While some might argue that medical genetics is emerging as a separate specialty and hence general practitioners may refer the concerned patients, the decentralized health-care system and socioeconomic disparity within the population of the country limit the access of a huge population to tertiary care hospitals and specialists [16], thus necessitating the need of general practitioners to be well-aware of medical genetics.

Moreover, with the advent of advanced research technologies, many diseases earlier solely attributed to environmental or lifestyle factors are now being reported to also have a genetic component to the same [17-19], further emphasizing the need to educate future health-care professionals about medical genetics, its impact on an individual's health, and the therapeutic requirements of such diseases. Existing evidence from a similar study conducted by Baars *et al*. in The Netherlands specifically, medical students nearing graduation also reported a limited knowledge about medical genetics [20]. Similar results were also recently reported by Alotaibi and Cordero from a study conducted with senior medical study students in the Kingdom of Saudi Arabia [21].

Assessing the awareness of medical students regarding the epidemiological and social impact of genetic diseases led us to identify that most medical students do not have very strong opinions in agreement or disagreement with the epidemiological and social impact of genetic diseases. Considering the median score of all respondents, the investigators identified that medical students agree to genetics being a major cause of infant mortality, any individual being susceptible to contracting genetic diseases, genetic disorders having a social stigma attached to them, and the necessity of including genetic education in school curricula. On the contrary, students have a neutral opinion on whether genetic disorders have a cure and disagree to hold parents accountable for a child getting genetic diseases. The lack of a strong opinion at all regarding the aforementioned facts about genetic diseases might be attributable to the limited knowledge of basic medical genetics. Among the others, what the investigators find most concerning is the student not having any opinion regarding curative therapy for genetic diseases. A clinician practicing health care in the community is expected to be able to address most if not all queries of patients and their guardians (in cases of the pediatric population) regarding the diseases, their therapeutic alternatives, and their prognosis. A neutral opinion regarding treatment alternatives/outcomes might not help the individual to satisfactorily address the query of the patient and/or his/her guardian. Therefore, it is crucial to augment the understanding of medical students so that they can cultivate accurate and evidence-based perspectives concerning clinical practice, particularly in the field of medical genetics.

Assessment of the attitude of medical students toward the occurrence and management of genetic disorders in society reported that medical students agreed to prevention is the best management of genetic disorders in our society and to post-marriage and pre-conception counseling to help prevent common genetic diseases in lower socioeconomic status. On the other hand, students did not have any opinion regarding the possibility of eradicating genetic diseases and disagreed with the social awareness lacking in our society. While the students had an overall optimistic attitude toward the management of genetic disorders in society, what concerned the investigators was the students' disagreement regarding the lack of social awareness regarding genetic disorders. As the current study itself identified the lack of knowledge about genetic disorders among medical students, it can be anticipated and has also been reported in the existing evidence [22,23] that the general population has very limited knowledge or social awareness about genetic disorders. Acknowledging this lack of awareness is essential to think toward addressing the same and eventually encouraging the prevention of genetic disorders through post-marriage and pre-conception counseling and genetic testing. Thus, the study also identified an

optimistic attitude for medical students toward the management of genetic disorders in society but also the need for encouraging a more practical approach toward the same.

The primary limitation of the study was a limited sample size. Surveyor Fatigue and the use of a web survey for data collection were identified as the primary reasons underlying the limited response rate. The study suggests that more evidence needs to be generated regarding the limited knowledge of medical students regarding medical genetics to support policies for inculcating formal genetic training in the medical curriculum. Furthermore, exposure to practical genetic practices - diagnostics, therapeutics, and counseling - is essential to improve student's attitudes toward genetic disease management.

CONCLUSION

The current study identified that medical students have limited knowledge about basic medical genetics, moderate awareness regarding the epidemiological and social impact of genetic diseases, and an optimistic attitude toward the management of genetic diseases in society. Curricular teaching and practical exposure might help increase knowledge and awareness and inculcate practicality in students' attitudes.

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Funding: None; Conflicts of Interest: None Stated.

How to cite this article: Johar P, Puri O, Verma PK. Assessment of knowledge, awareness, and attitude toward genetic diseases among medical students studying in a tertiary health-care teaching hospital in Uttarakhand, India. Indian J Child Health. 2023; 10(7):85-89.