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ORIGINAL ARTICLE

Assessing the knowledge and awareness of the Taif community about genetic diseases

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ABSTRACT

Background: Diseases have a genetic basis, wherein changes in the human deoxyribonucleic acid and variances in its activities, which the environment may influence, contribute to disease processes. The risk of developing a disease is higher for people with genetic susceptibility when other risk factors such as lifestyle and environmental factors are present besides genetic change. The Taif community's attitudes toward genomes and their awareness are not well documented. The study aimed to assess the knowledge and awareness about genomes among Taif residents.

Methods: A cross-sectional study was conducted on 361 participants residing in Taif city, Saudi Arabia, using a pretested questionnaire. The first part of the questionnaire collected participants' sociodemographic information and the second part measured the awareness and knowledge related to genetics and diseases.

Results: All responses were subjected to statistical analysis. Among the studied subjects, 6.6% of the participants demonstrated good knowledge of genetics and diseases and 45.7% had poor knowledge. 95.6% were aware of health problems caused by a combined effect of genetics, environment, and lifestyle; 83.7% agreed that genes play a role in disease processes; and 92.8% agreed that individuals with a family history of a particular disease could benefit from undergoing a genetic test for that disease.

Conclusion: The study participants had inadequate knowledge, suggesting that more effort is required to educate them about the advantages and limitations of genetic testing on a social and personal level to ensure that people make well-informed decisions. The Saudi genome program is one among such programs targeting at educating the community.

Keywords: Genome, awareness, Taif community, genetic diseases, knowledge.

Introduction

All living things contain a complete set of genes made up of deoxyribonucleic acid (DNA) segments. The human genome consists of 3.2 billion DNA bases (1). Our genome is approximately 99.9% identical to every other human being. It is the 0.1% variation that is of interest in healthcare as understanding this variation can help in the prediction, prevention, diagnosis, and treatment of disease (1). Therefore, the genetic similarity between siblings can range from 0% to 100%. Identical twins share 100% of their DNA. On average, each individual is genetically close to both parents and siblings (2). Genetic diseases occur when genes are mutated. Gene mutation refers to the alteration of genes or chromosomes. These alterations could be in the number or structure of the chromosome. Gene mutations may be hereditary, wherein the parents inherited the alteration. The disease appears at birth or is acquired, which means that the alteration happens during the individual's lifetime, like exposure to sunlight leading to skin cancer (3). Although there are many possible risk factors for human disease, genetic background is often among the strongest risk factors for common disease complexes, such as congenital heart disease (4,5), diabetes (5,6),

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hypertension (5,7), asthma (5,8), and cancer (9). Replacement of a single base pair is the most common cause of DNA variation; when these variants occur at a population frequency of 1% or more, they are referred to as single nucleotide polymorphisms (10).

The gene-environment study represents a broad class of genetic association studies focused on understanding how differential responses to environmental exposures and differential effects on variations in other genes are associated with human genetic variability (11). To explain the concept of gene-environment interactions, recent studies describe the genetic mutations associated with the differential reaction to cigarette smoke and its association with lung cancer (11). Ethnicity is the product of several factors, including social and political pressures, history, faith, and nationality. Understanding a population's ethnic mix can facilitate healthcare delivery by helping to target services such as screening initiatives, education, and resource distribution (12). Most people have a history of at least one chronic disease in their families. If an individual has a close family member with a chronic disease, they may be more likely to develop the disease, especially if more than one close relative has the disease or a family member developed the disease at a younger age than usual; however, when the relatives develop the disease in old age, it is considered acquired (13). Gene mutations are one of the risk factors that may lead to cancer development; screening programs and prevention can reduce mortality from cancer (14). Therefore, by carrying out a genetic mutation test, we can detect mutations earlier, and intervention becomes easier, more useful, and effective. Genetic testing is a medical test that identifies changes in chromosomes, genes, or proteins. It can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder (15). Not everyone with a family history of a disease may benefit from undergoing a genetic test because genetic testing cannot completely predict the future and has limited scope, providing only restricted results about the genetic condition (16).

Pharmacogenetics is a component of personalized medicine involving the study of genetic differences among individuals that cause varied responses to a drug to improve the effectiveness and safety of drugs (17). Personalized medicine has shown great potential, with a promising future hoping to increase the effectiveness of medical treatment (17). Personalized medicine focuses on studying the genetic background of each patient, which helps in understanding the disease and choosing the best treatment for each patient instead of treating all patients the same (18).

The historically important summer capital of the Kingdom of Saudi Arabia (KSA) is Taif, located in the Makkah region. In 2010, 580,000 people were living in the city. Over 1 million people live in the city today (19). A majority of its residents are younger than 30 years old (19). Several sociodemographic characteristics, including age, gender, ethnicity, and education level, are considered sociodemographics in our study. Here, we are concerned with assessing and shedding light on the knowledge and awareness of the Taif community about the genome.

Subjects and Methods

This cross-sectional study was conducted during the period from April to August 2021 to assess the awareness and perception of genomic medicine in the KSA. Ethical approval for the study was obtained from the Ethical Committee of Health Affairs of Taif city. All adults of both genders who lived in Taif city in KSA and agreed to answer the questionnaire were included in the study. However, individuals who did not live in Taif and those who refused to complete or did not complete the questionnaire were excluded. A questionnaire regarding the community's awareness and perceptions of genomes (20) was used, and information was collected from 361 random participants. The questionnaire included items on demographics (e.g., age, occupation, nationality, and education), assessed community members' awareness of genomic medicine, their knowledge of genetics and genetic testing, moral attitudes, confidence in regulatory agencies, the anticipated uses of technology and expectations regarding future technological advances. It included 14 true/false knowledge statements. Most statements fell into four broad categories: (1) family history and inheritance, (2) screening for common diseases, (3) genetic testing, and (4) laws governing the use of genetic information. The final survey consisted of 16 true/false statements. Data were entered using Microsoft Excel version 16.16.23, and statistical analysis was carried out using Statistical Package for the Social Sciences version 23 (International Business Machines Corporation Corp., USA) by an independent biostatistician. Categorical variables were analyzed using Pearson's chi-square test. Continuous variables were expressed as mean and standard deviation. A significant value (p-value) ≤ 0.05 was considered statistically significant.

Results

Sociodemographic characteristics

The study assessed the knowledge and awareness of the relationship of genes with various diseases. It included responses from 361 participants from the Taif region, of which 60.1% were women, and the majority (91.4%) was of Arabic origin. The age distribution showed that 30.7% was ≤29 years, and 59.6% had graduate-level education, as shown in Table 1. The correct and wrong responses to each question are given in Table 2.

Correlation of genetics with different variables

Environment

It was found that 95.6% of the participants agreed that most health problems are caused by genetics, environment, and lifestyle. This response did not show any statistical association with age, gender, and education level ($p \geq 0.05$).

Table 1. *Sociodemographic characteristics.*

		Frequency	Percentage %
Gender	Female	217	60.1
	Male	144	39.9
Ethnicity	Arab	330	91.4
	Asian	23	6.4
	African	1	0.3
	Other	7	1.9
Age (in years)	≤29	111	30.7
	30-39	84	23.3
	40-49	82	22.7
	50-59	64	17.7
	60-69	19	5.3
	≥70	1	0.3
Educational level	Primary	2	0.6
	Secondary	92	25.5
	Graduate level	215	59.6
	Postgraduate level	33	9.1
	PhD	19	5.3

Table 2. *Community's awareness and perceptions of genomes (20).*

	Responses	
	Wrong (*n*, %)	Correct (*n*, %)
Most health problems are caused by a combination of genetics, environment, and lifestyle.	16 (4.4%)	345 (95.6%)
Cancer screening is only recommended for people with a family history of cancer.	109 (30.2%)	252 (69.8%)
If you have a family history of a disease, you are more likely to develop it.	112 (31%)	249 (69%)
Everyone has genetic variations that make them more susceptible to certain diseases.	61 (16.9%)	300 (83.1%)
A person's race and ethnicity can influence how likely they contract the disease.	101 (28%)	260 (72%)
It is important to know how old your relatives were when they contracted the disease.	109 (30.2%)	252 (69.8%)
If you have some variation in a gene that causes cancer, there is nothing you can do to prevent cancer.	120 (33.2%)	241 (66.8%)
Breast cancer can only be inherited from the mother's family.	99 (27.4%)	262 (72.6%)
Genes play a part in almost all diseases.	59 (16.3%)	302 (83.7%)
Genetic testing can be done to see how a person's body reacts to certain medications.	19 (5.3%)	342 (94.7%)
People are more genetically similar to their parents than they are to their siblings.	118 (32.7%)	243 (67.3%)
It is possible to do genetic testing for most common diseases such as heart disease, diabetes, high blood pressure, and others.	66 (18.3%)	295 (81.7%)
All women will benefit from having a genetic test for breast cancer.	50 (13.9%)	311 (86.1%)
Everyone with a family history of a particular disease can benefit from having a genetic test for that disease.	26 (7.2%)	335 (92.8%)

Ethnicity

It was agreed by 72% of the participants that race or ethnicity has a relationship with some diseases, and a higher percentage of correct answers was observed in participants aged ≤29 years ($p = 0.001$; Figure 1).

Relationship of the genetics with diseases

Genetic variations

Among the study subjects, 83.1% of the participants agree that everyone has genetic variations that make them more susceptible to certain diseases. This response was not statistically associated with age, gender, and education level ($p \geq 0.05$).

Diseases

Among the study subjects, 83.7% of participants stated that genes play a significant role in almost all diseases, and this correct response was more frequently given by participants with postgraduate- and PhD-level education ($p = 0.004$; Figure 2).

Gene inheritance

Family history

The percentage of participants who believed that people who have a family history of a disease are more likely to develop the disease was 69%; this response was statistically and significantly associated with gender, wherein women gave the correct response more frequently than men ($p = 0.016$; Figure 3).

Genetic similarity

About 67% of the participants believed that people are more genetically similar to their parents than their siblings, and no statistically significant relationship with age, gender, and education level was seen ($p \geq 0.05$).

Inherited and acquired

It was seen that 69.8% of the participants believed that it is important to know how old their relatives were when they contracted a disease; this response did not have any statistically significant association with age, gender and education level ($p \geq 0.05$).

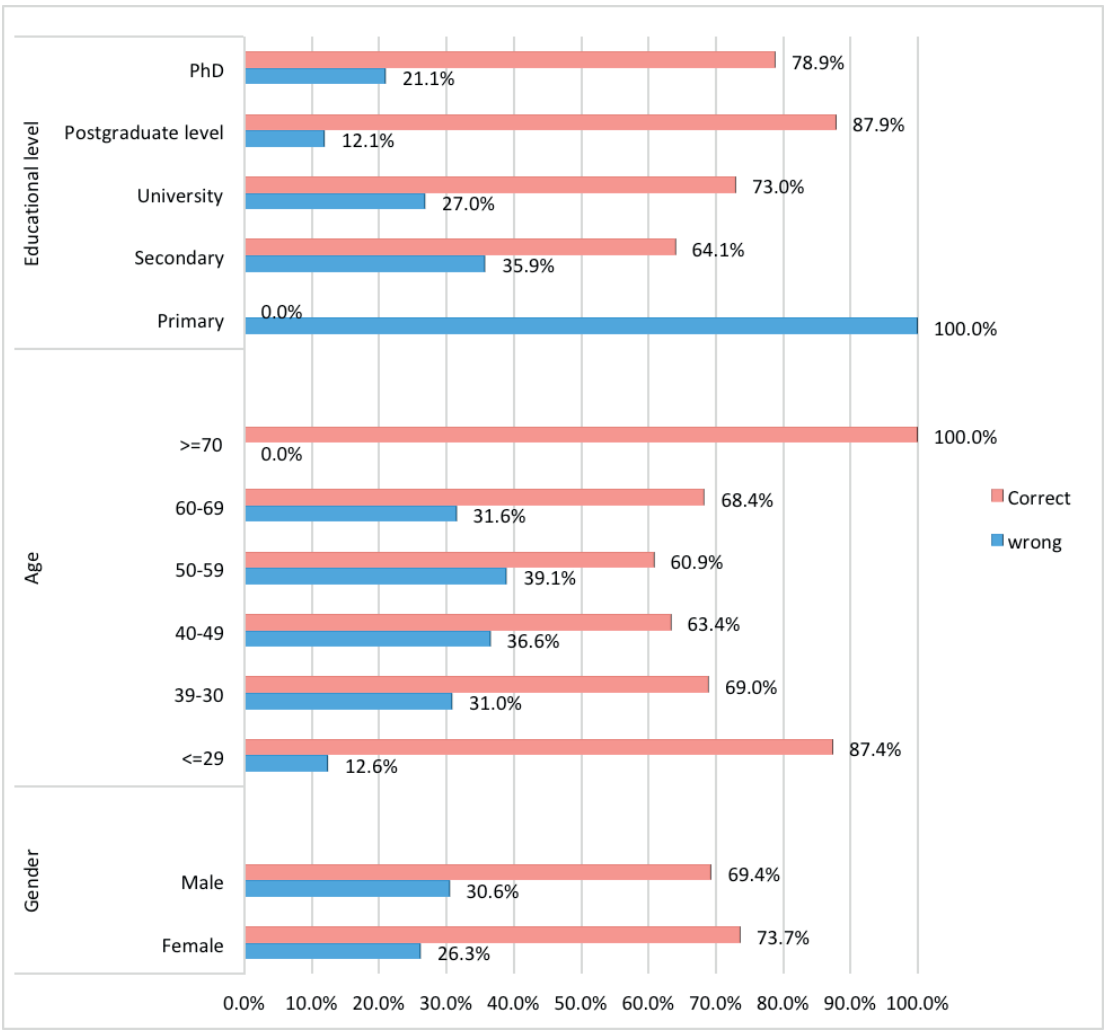

Figure 1. *Person's race and ethnicity, representing the participants' responses to whether the person's race and ethnicity can influence how likely they are to contract a disease.*

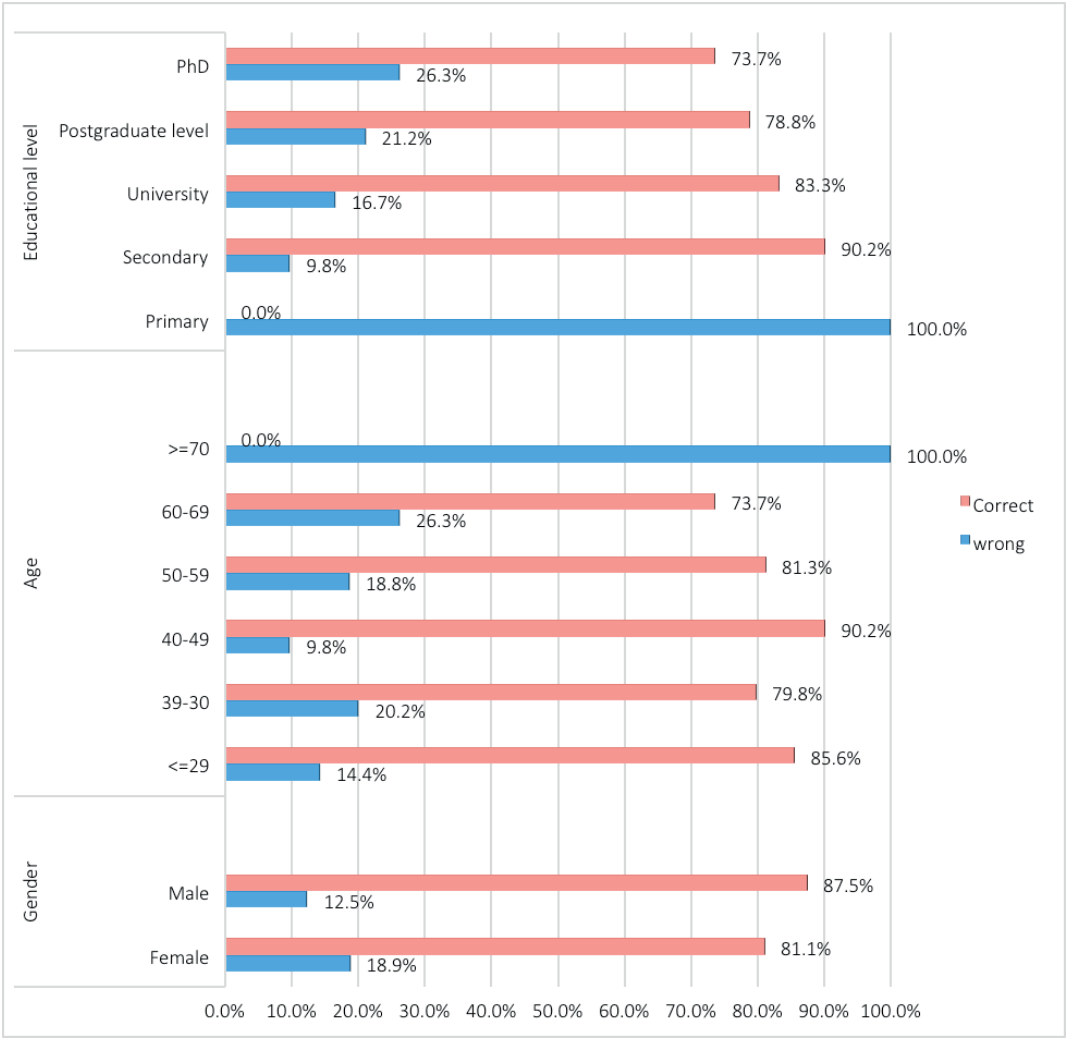

Figure 2. *Genes and diseases, showing the participants' response to whether genes play a part in almost all diseases.*

Cancer

Most of the participants (72.6%) also believed that breast cancer could not only be inherited from the mother's side of the family but also has paternal genetic susceptibility, a response that did not statistically and significantly associate with age, gender, and education level ($p \geq 0.05$).

Genetic testing

Family history

Among the study subjects, 92.8% of the participants believed that everyone with a family history of a particular disease could benefit from undergoing a genetic test for that disease. However, this response did not show a statistically significant relationship with age, gender, and education level ($p \geq 0.05$).

Common diseases

Approximately 82% believed that it is possible to carry out genetic testing for most common diseases, such as heart disease, diabetes, high blood pressure, and others; this response had no statistically significant relationship with age, gender, and education level ($p \geq 0.05$).

General information about genetics

Cancer screening

Majority of the participants (86.1%) believed that all women would benefit from undergoing a genetic test for breast cancer, with comparatively more reports among participants with education at the graduate level or higher ($p = 0.036$). Of the participants, 69.7% believed that cancer screening is not recommended only for people with a family history of cancer; this response was more frequent among participants with a comparatively high education level ($p = 0.009$; Figure 4).

Prevention

It was believed by 33.2% of the participants that any measures could not prevent diseases with a genetic predisposition. This belief was not statistically associated with age, gender, and education level ($p \geq 0.05$).

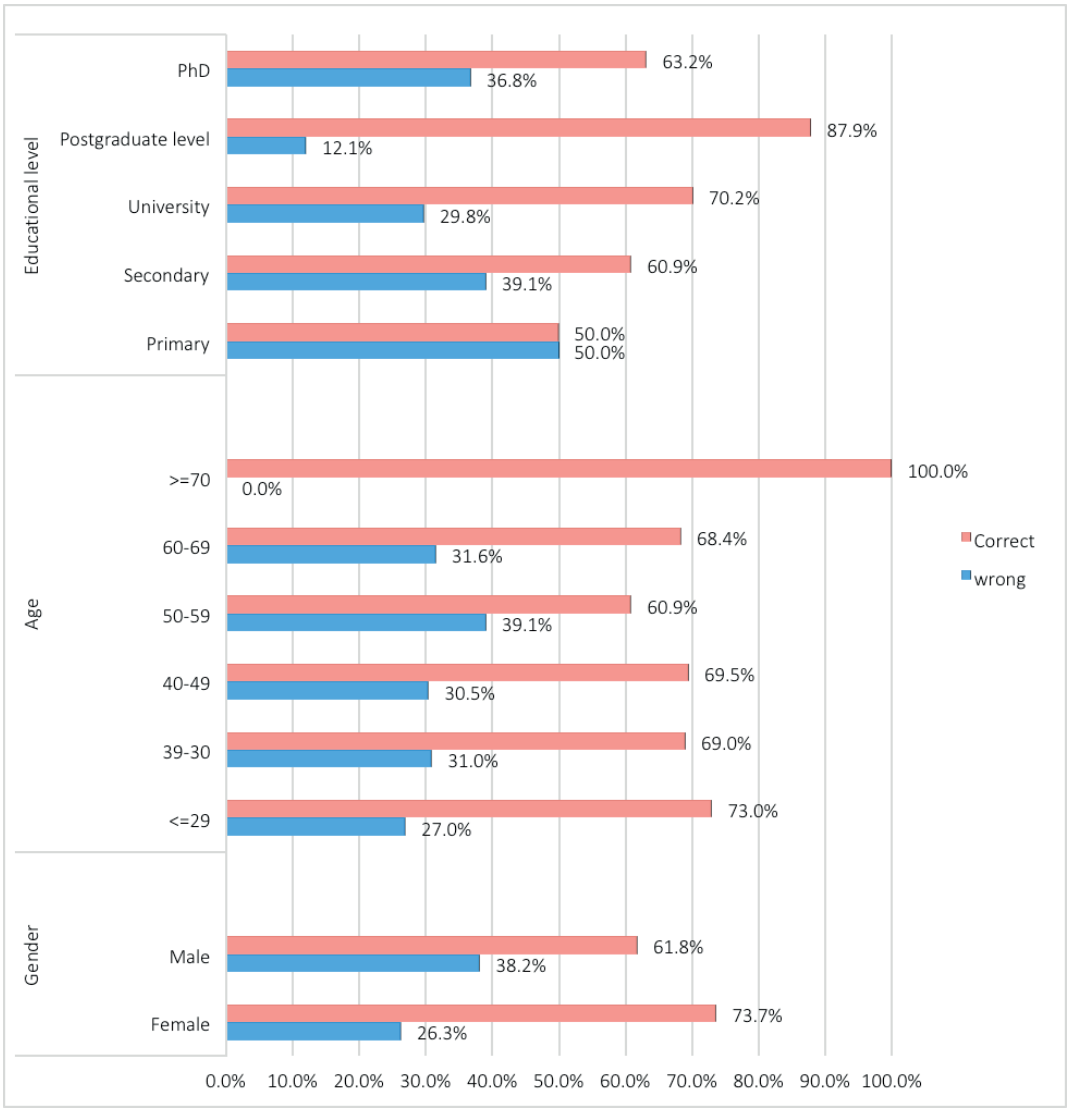

Figure 3. *Family history of a disease, showing that the contributors' reaction to the family history of disease makes the person more likely to develop it.*

Personalized medicine

Majority of the participants (94.7%) had the opinion that genetic testing (screening) can be carried out to evaluate the body's reaction to certain medications, and this response did not show any statistical significance concerning age, gender, and education level ($p \geq 0.05$).

Knowledge level

To calculate the knowledge level of the participants, their responses were checked, and a score of 1 was given for correct responses, while no score (0) was given for wrong responses. The maximum score a participant could get was 14 and the minimum was 0. The obtained scores were then converted into percentages. Those who achieved a score ≥75% were categorized as having "good" knowledge, those with 60%-75% as "fair," and those with ≤60% as "poor." The analysis showed that only 6.6% (n = 24) of the participants had good knowledge and 45.7% had poor knowledge (Figure 5).

When the knowledge levels of the participants were compared based on gender, no statistically significant differences were observed (p = 0.974). More participants of Asian origin were found to have "good" knowledge levels compared with others, but this difference was not statistically significant (p = 0.069). Comparing knowledge levels by participant age showed that more participants aged 50-59 years and ≤29 years demonstrated "good" knowledge levels, with a statistically significant association (p = 0.017). However, no statistically significant difference was observed in the knowledge levels of participants according to their highest educational qualification (p = 0.069; Table 3).

Discussion

Genomic research has revealed many associations between specific genetic variants and disease outcomes (21). In the past, genomic research was concentrated around rare and single-gene disorders, in which the mutation of a single gene was responsible for

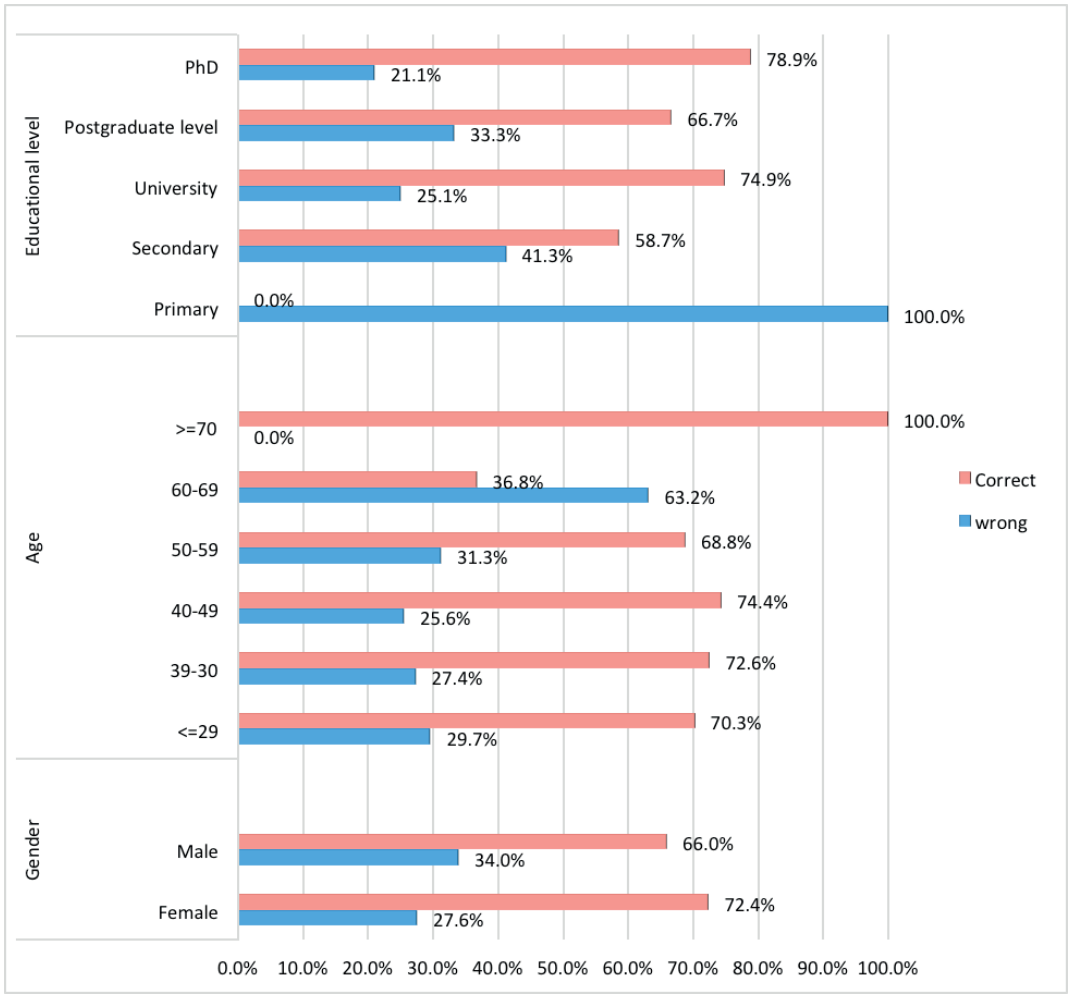

Figure 4. *Cancer screening. Response to whether cancer screening is only recommended for people with a family history of cancer.*

disease occurrence. Diseases such as cystic fibrosis, Duchenne muscular dystrophy, fragile X syndrome, and Huntington's are caused by the mutation of specific genes (22,23). Genetic studies have shifted their focus in recent years and are paying more attention to common and complex diseases such as cardiovascular diseases, hypertension, cancer, diabetes, and asthma (24). Multiple genetic and environmental factors are associated with these common diseases; thus, it is challenging to study their genetic relationships, unlike other rare diseases usually caused by a single gene disorder (25). Genes play a significant role in many common diseases that are leading causes of death, such as cardiovascular diseases and cancer (26). Therefore, such genetic studies raise concerns in communicating with patients and the public about genetic contributions to these diseases when passing on disease risk messages.

More than half of the participants are female, aged below 29 years, and they are at the graduate level. The sociodemographic result shows that most of the participants are from the college category, which is easily accessed to spread this questionnaire.

It is a well-known fact that many diseases are caused by the interaction of genetic and environmental factors, but the fundamental concept is unresolved. Diseases

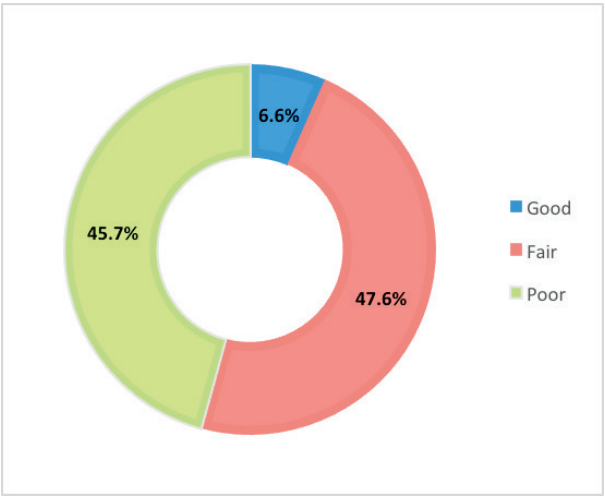

Figure 5. *The knowledge level of the participants, where the total number of participants is 361.*

such as sickle cell alpha-thalassemia, cystic fibrosis, and spinal muscular atrophy are predominantly found in certain ethnic groups. Many of these diseases are linked to rare mutations of a single gene (27,28). Diseases are frequently considered to be genetically inherited because they run in families. However, it should be made clear that such patterns are not only due to genetic susceptibility

Table 3. *Relationship of knowledge level with sociodemographics.*

		Knowledge level			Total	*p*-value
		Good	Fair	Poor		
Gender	Female	14	103	100	217	0.974
		6.5%	47.5%	46.1%	60.1%	
	Male	10	69	65	144	
		6.9%	47.9%	45.1%	39.9%	
Ethnicity	Arab	21	163	146	330	0.069
		6.4%	49.4%	44.2%	91.4%	
	Asian	3	9	11	23	
		13.0%	39.1%	47.8%	6.4%	
	African	0	0	1	1	
		0.0%	0.0%	100.0%	0.3%	
	Other	0	0	7	7	
		0.0%	0.0%	100.0%	1.9%	
Age	≤29	11	63	37	111	0.017
		9.9%	56.8%	33.3%	30.7%	
	39-30	2	45	37	84	
		2.4%	53.6%	44.0%	23.3%	
	40-49	4	32	46	82	
		4.9%	39.0%	56.1%	22.7%	
	50-59	7	24	33	64	
		10.9%	37.5%	51.6%	17.7%	
	60-69	0	7	12	19	
		0.0%	36.8%	63.2%	5.3%	
	≥70	0	1	0	1	
		0.0%	100.0%	0.0%	0.3%	
Educational level	Primary	0	0	2	2	0.069
		0.0%	0.0%	100.0%	0.6%	
	Secondary	3	36	53	92	
		3.3%	39.1%	57.6%	25.5%	
	University	20	108	87	215	
		9.3%	50.2%	40.5%	59.6%	
	Postgraduate level	1	18	14	33	
		3.0%	54.5%	42.4%	9.1%	
	PhD	0	10	9	19	
		0.0%	52.6%	47.4%	5.3%	

but are more commonly caused by a shared environment and lifestyle (29).

The findings of our study show that nearly three-quarter of the participants believed that a person's race or ethnicity could influence how likely they are to contract a disease. The health ministry is already running an awareness campaign for pre-marriage tests to examine the risk of having children affected with the most common genetic diseases in Saudi Arabia like beta-thalassemia, which may explain the high awareness response about ethnicity.

The findings of our study show that knowledge regarding genetic contributions to diseases was fair, with only a small proportion of participants demonstrating good knowledge. Despite traditional demographic predictors, such as ethnicity and age, education level differs in awareness and attitudes. Particularly when it comes to genetics, sociocultural factors may affect public awareness. However, most of the participants believed that there is a genetic contribution to every disease. This could be due to higher reporting of genetic diseases and the permeation of genomic research into our culture, thus increasing public awareness (30).

Furthermore, approximately 70% of the participants believed that cancer screening is only recommended

for people with a family history of cancer in this study. It is well-established that cancers like breast cancer, colorectal cancer, and ovarian cancer occur more frequently in individuals with a family history of these diseases (31,32). Early screening in patients with a family history would help in early detection and has the potential to create awareness and/or motivate the adoption of risk-specific strategies to decrease the disease burden in the community. It is recommended that people with hereditary cancer susceptibility undergo genetic counseling, and early detection protocols for different types of cancers vary according to familial risk (33,34).

Molecular screening tests are currently available to identify diseases at early stages and can be used as a key component of disease prevention strategies (35).

It is essential to understand that not all individuals respond to medicine in the same beneficial way. Garrod first proposed this in 1923, and more recently, the term "pharmacogenomics" has been used to convey that drug response variability may be found in certain individuals or across a population (36). Evidence shows that only half of all patients respond positively to a drug. Thus, the other half may experience therapeutic delays or may not be properly medicated due to a lack of drug efficacy (37).

Genomic variations can affect drug metabolism, transport, and targets (38). Hence, it is important for physicians and pharmacists to explain this drug-response variability to patients when prescribing and dispensing medications. It has been shown that physicians lack the training to apply knowledge of this drug-response variability in clinical decision-making (39). In our study, most participants agreed that genetic testing could be conducted to see how a person's body reacts to certain medications.

In KSA, most of the population uses mass media for health communication and as primary health and science information sources. It is important to translate genetic research findings to the public to establish public acceptance of the role of genetics in many diseases, especially in chronic diseases such as diabetes, cardiovascular diseases, cancer, obesity, and hyperlipidemia. Social media campaigns should not only make efforts to encourage the public to be aware of genetic susceptibility in chronic diseases but also simultaneously focus on discussing the role of environmental and lifestyle factors in disease causation. Understanding people's awareness, knowledge, beliefs, and attitudes could reduce gaps in knowledge and concerns by exposing misunderstandings in genetic research. Thus, intervention efforts at the individual and population levels are needed to improve awareness, giving special attention to subgroups of lower socioeconomic status and minorities. Our research findings should be interpreted considering specific limitations. First, knowledge and awareness were recorded based on self-reports, which could have resulted in social desirability bias as these are subject to individual interpretation. Second, we did not assess the source of information and, therefore, cannot ascertain which factors influenced the awareness level of the participants.

Conclusion

We found an imbalance between knowledge and awareness despite greater awareness, with nearly half of the participants demonstrating poor knowledge. More effort is required to educate the public about the benefits, demerits, and dangers for informed decision-making and a proper understanding of genomic risk. Longitudinal studies are needed to evaluate whether providing genetic information to previously ignorant people helps convey genomic information in a manner that promotes good results while minimizing negative consequences.

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Conflict of interest
The authors declare that there is no conflict of interest regarding the publication of this article.

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Consent to participate
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Ethical approval
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