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ORIGINAL ARTICLE

Knowledge and awareness of genetic diseases among residents of the western region of Saudi Arabia

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ABSTRACT

Background: Genetic diseases result from gene mutations that disrupt the function of body systems, and most of them cannot be treated. Genetic diseases are a serious issue responsible for severe economic, emotional, and health impacts on patients and their families. In Saudi Arabia, consanguineous marriage is common, leading to an increased prevalence of genetic diseases. Therefore, Saudi Arabia has endeavored to reduce the frequency of genetic diseases by implementing the premarital screening program, which is mandatory and accessible for people planning to get married, and the genetic counseling program.

Methods: In this study, we investigated the awareness and knowledge of genetic diseases among residents of the western region of Saudi Arabia, and we conducted a cross-sectional survey focusing on the level of awareness and knowledge of genetic diseases and available health care.

Results: We found that the residents of the western region had good knowledge about genetic diseases, but 96.10% lacked the awareness of health services provided for this type of disease. Furthermore, 91.90% of them had a positive attitude toward genetic testing.

Conclusion: The present study found that Saudi society had a good attitude toward genetic testing, such as prenatal and preimplantation genetic diagnosis. Many of the study participants were not aware, where to access genetic tests for their families, and most considered health services for patients with genetic diseases to be rudimentary, thus pointing out a lack of awareness and knowledge toward health services.

Keywords: Genetic diseases, awareness and knowledge, western region, Saudi Arabia, mutations, Genetic testing.

Introduction

Genetic diseases arise as a result of mutations or abnormalities in the DNA sequence (1,2). The typical assumption among laypeople is that only heritable diseases have a genetic basis, for example, cystic fibrosis, phenylketonuria, and some cancers (1). However, an individual's genetic makeup can determine whether or not a gene is expressed correctly, consequently resulting in disease. For example, an alteration to a gene involved in the immune system could increase susceptibility to infection compared to an individual carrying the wild-type version of the same gene. Understanding the human genome and its variations in the human population are, therefore, integral to understand and treat diseases and can provide a foundation for improved treatments and preventative measures (1). In Saudi Arabia, there are many genetic and chromosomal disorders such as Down syndrome, congenital malformations, congenital heart disease (CHD), cystic fibrosis, Duchene muscular dystrophy (DMD), hereditary recessive deafness, hereditary blindness and visual impairment, thalassemia,

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and sickle cell disease (SCD) (3). Furthermore, El Mouzan et al. (4) identified several genetic disorders to be common in the population. For example, Down syndrome occurred at a frequency of 1.8 in every 1,000 live births, and SCD was reported to be endemic in some areas of the country, with a prevalence ranging from 91 to 99 per 10,000 live births in the Eastern Province. In particular, the eastern Saudi cities of Riyadh and Dammam had SCD prevalence rates of 51.3% and 52.0%, respectively. Other genetic diseases, such as glucose-6-phosphate dehydrogenase deficiency, beta-thalassemia, and CHD, were also common (4). The study conducted by El Mouzan et al. (4) demonstrated that consanguinity is a significant cause of many genetic diseases, particularly CHD. Moreover, consanguineous marriage is common in Saudi Arabia, occurring in 57% of marriages, leading to an increased prevalence of genetic diseases (5-7).

In past decades, Saudi Arabia has endeavored to reduce the frequency of genetic diseases through the use of national, communal, awareness, and preventive programs such as the premarital screening program, which is mandatory and free of cost for people who are planning to get married, and the genetic counseling program (5,6). Although the premarital screening program has been successful, it only tests for two genetic diseases, such as beta-thalassemia and SCD. This is a significant flaw in the program as several research studies have identified many other genetic diseases to be common in Saudi Arabia (5). Genetic diseases can impact individuals socially and psychologically as well as medically (7-10). Therefore, in this study, we conducted a cross-sectional survey focusing on the level of awareness and knowledge of genetic diseases and available health care among residents of the western region of Saudi Arabia. The survey was published on social media and distributed among randomly selected individuals.

Subjects and Methods

We conducted a cross-sectional study using an electronically distributed survey. The survey was created using Google Forms software, translated into Arabic, and distributed through social media channels (WhatsApp and Twitter) to residents of the western region of Saudi Arabia. All surveys were anonymous, with no identifiers used to ensure confidentiality of all participants' information. The participants were invited to participate in the study by a cover letter explaining the importance of their contribution to this study. Survey participants were included or excluded from the study based on predetermined criteria, and the total number of included participants was 433. The survey had four sections: a collection of demographic data and surveys of the participants' knowledge of genetic diseases, diagnosis and prevention methods, and where to obtain medical information and diagnosis of genetic disorders in the western region of Saudi Arabia. All responses were transferred to Microsoft Excel sheets for statistical analysis. All data were categorized and analyzed using descriptive statistics such as percentages, and all statistical figures were built by Microsoft Excel tools.

Results

Participant demographics

Public knowledge and awareness regarding genetic diseases are crucial for effective treatment implementation. Therefore, the questionnaire was designed to investigate this in the western region of Saudi Arabia. A total of 433 participants were completed the questionnaire (Table 1). About 57.8% of participants were between the ages of 20 and 35 (Table 1 and Figure 1); the majority of participants were male (239 [55.8%]), and 189 participants were female (32.8%). In addition, more than 80% of participants held a university level degree (bachelor's degree). Finally, we investigated the location of participants, and the most common cities were Jeddah (32.3%), Makkah (27.9%), Taif (12.2%), Al-Madina Al-Munawarah (7.6%), and Yanbu (1.6%). The remaining 18.4% of participants were located in other minor cities in western Saudi Arabia (Figure 1). A detailed breakdown of the demographic profile of the study respondents is shown in Table 1.

Knowledge of genetic diseases among survey participants

About 57% of participants did not know about the prevalence of genetic diseases in the western region of Saudi Arabia (Table 2). However, the majority (72.2%) of participants chose consanguinity as the cause of genetic disease prevalence, compared to 38.6% and 22.4% who chose a lack of health awareness or mutations, respectively. Furthermore, 65.1% answered in the affirmative when asked whether consanguinity was the primary cause of genetic disease in the western region. Most of the

Table 1. *Demographic characteristics of the participants: age, gender, education level, and city.*

Item	Category	%
Age	10-15	0
	16-20	9.20
	21-25	29.60
	26-35	28.20
	Above 36	32.80
Gender	Male	55.80
	Female	44.20
Education level	Middle school	0.70
	High school	12
	University	80.10
	Postgraduate	7.20
City	Jeddah	32.30
	Makkah	27.90
	Taif	12.20
	Madinah	7.60
	Yanbu	1.60
	Others	18.40

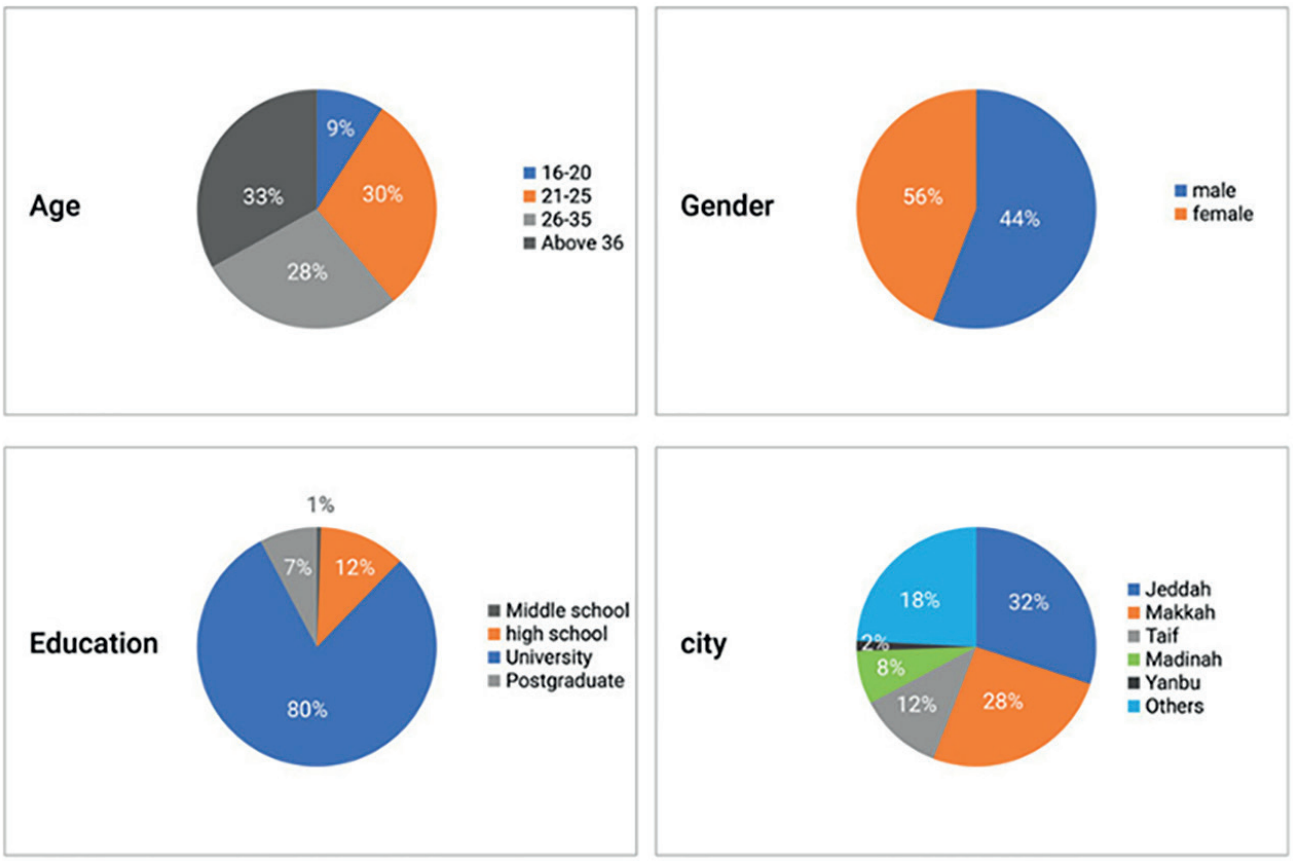

Figure 1. *Demographic characteristics of the participants: age, gender, education level, and city.*

Table 2. *Participants' knowledge about genetic diseases.*

Survey questions	Responses	%
In your opinion, how prevalent genetic diseases are in the western region?	Prevalent	27.50
	Not prevalent	15.50
	Do not know	57
What are the reasons for genetic diseases prevalence in the western region?	Consanguinity	72.5
	Mutations	22.40
	Lack of health awareness	38.60
	others	3.30
Do you believe that consanguinity is a major reason for genetic disease?	Yes	65.10
	No	6
	Maybe	28.90
In your opinion, do you think genetic diseases are infectious?	Yes	3.50
	No	86.10
	Maybe	10.40
Do you know what the most common genetic diseases in Saudi Arabia are?	Yes	31.90
	No	69.10
Do you know how to reduce the risk of having a genetic disease?	Yes	45.50
	No	54.50
Do you have any information on how genetic diseases are diagnosed?	Yes	33
	No	67
Is there a possibility of having normal children for parents who suffer from a genetic disease?	Yes	55.20
	No	7.40
	Maybe	37.40

participants (86.1%) knew that genetic diseases are not infectious. Still, nearly 70% of participants did not know what the most common genetic diseases in Saudi Arabia, and 67% had no knowledge of how genetic diseases are diagnosed. Furthermore, only 55.2% of respondents thought that parents who suffer from a genetic disease could have healthy children.

Awareness regarding genetic diseases among survey participants

In this section of the survey, we investigated the level of awareness that the participants had regarding genetic diseases and how they obtained health information and advice about these diseases (Table 3 and Figure 2). Nearly 53.3% of participants considered themselves to have a medium level of awareness, and 38.6% considered their awareness level below. Participants reported websites to be the most common resource for obtaining information regarding genetic diseases (39.5%), followed by social media (27.3%); 18.2% used other unspecified sources. Concerningly, only 15% of participants reported specialist doctors to be their most common source of information (Table 3 and Figure 2A), and 74.1% of participants had never seen an awareness campaign or received information through text messages regarding the health risks of genetic diseases.

Finally, we asked participants about their opinion on the best way to protect the community from the consequences of genetic diseases. The majority of participants (73.2%) chose premarital testing as the best option. An increase in health awareness was selected as the second-best option by 66.5% of respondents (Table 3 and Figure 2B). In addition, 41.6% and 46.6% of respondents believed that launching awareness campaigns in educational institutions and increasing genetic disease testing in newborns would help increase protection against genetic disorders, respectively (Figure 2B).

Awareness of health service for genetic diseases among survey participants

In this part of the survey, we included questions assessing the participants' awareness of available health services

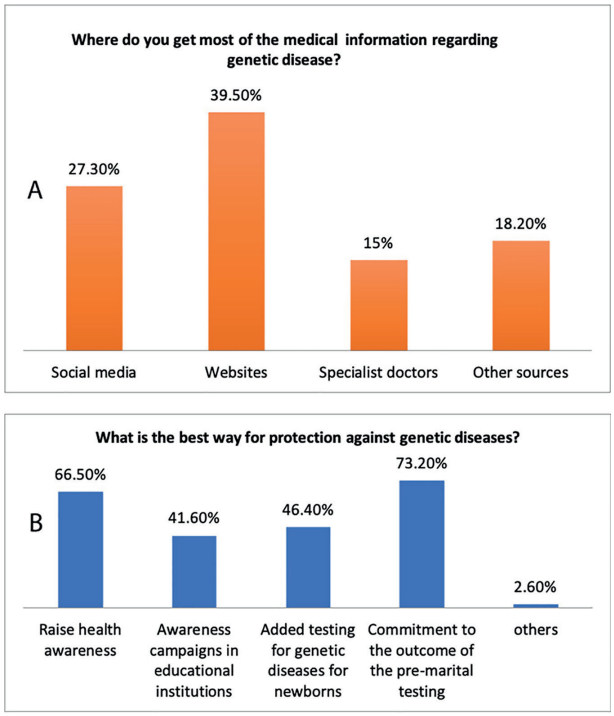

Figure 2. *Participants' awareness regarding genetic diseases. (A) The participants' answers about where they get medical information. (B) The best way of genetic prevention diseases based on the participants' responses.*

Table 3. *Participants' awareness regarding genetic diseases.*

Survey questions	Responses	%
What is your level of awareness regarding genetic diseases and associated risks?	High	8.10
	Medium	53.30
	Low	38.60
Do you have a family member that suffers from a genetic disease?	Yes	32.80
	No	67.20
Where do you get most of the medical information regarding genetic diseases?	Social media	27.30
	Websites	39.50
	Specialist doctors	15
	Other sources	18.20
Have you ever encountered an awareness campaign or received a text message regarding awareness about genetic diseases?	Yes	25.90
	No	74.10
What is the best method of protection against genetic diseases? Multiple answers can be selected.	Raise health awareness	66.50
	Awareness campaigns in educational institutions	41.60
	Added testing for genetic diseases for newborns	46.40
	Commitment to the outcome of premarital testing	73.20
	Other	2.60

for genetic diseases (Table S1 and Figure 3). The majority of participants (60.3%) reported that they did not know where they could be assessed for genetic diseases (Figure 3D). Furthermore, 66.1% did not know what actions to take if they have concerned about their children being affected by genetic diseases (Figure 3E). Almost all participants (91.9%) agreed to add more tests for genetic diseases in premarital testing programs (Figure 3A). When asked about genetic counseling, 96.1% of participants have never contacted a genetic counselor, and 64.2% did not know if genetic counseling was available in their city or town (Figure 3B and C). Overall, most of the respondents (68.6%) considered the available health services for patients with genetic diseases to be inadequate (Figure 3F).

Discussion

Genetic diseases are a serious issue responsible for severe economic, emotional, and health impacts on patients. Several studies showed that patients with genetic diseases need more regular visits to clinics and stay longer in hospitals, which negatively impact the healthcare cost and economy (11,12). In terms of emotional impact, genetic diseases can negatively affect both patients and their caregivers, with much-experiencing anxiety, guilt, anger, uncertainty, sorrow, and severe depression (5). Health impacts vary between diseases. For patients with sickle cell anemia, the production of abnormal red blood cells gives rise to blood clots, consequently impacting oxygen transport (5,8). Awareness of the causes of genetic diseases and knowledge of possible preventative measures are crucial to reduce the prevalence of these diseases and the high costs of health care. In this study, we investigated the level of knowledge and public awareness of genetic diseases among residents of the western region of Saudi Arabia. A significant finding was that while most participants understood the most common cause of hereditary diseases to be inbreeding, respondents with minimal educational levels did not have sufficient knowledge of the severity of genetic diseases and the possible health, emotional, and societal impacts. Moreover, most of the participants had never encountered any campaigns aiming to raise awareness of genetic diseases or received text messages regarding genetic diseases. Encouragingly, the general attitude toward increasing awareness of the causes of genetic diseases was positive.

The credibility of medical information is crucial for raising awareness of genetic diseases. Here, we found that most of the participants obtained medical data from websites instead of contacting specialist doctors. In addition, many respondents admitted to use social media for information regarding genetic disorders, indicating that websites and social media can be powerful tools for increasing public knowledge of genetic diseases. Moreover, a previous study identified social media and television as the most effective platforms for improving health awareness in many developed countries (13). Utilizing the media to raise public awareness of hereditary diseases in Saudi Arabia is likely to affect the overall knowledge people have of genetic diseases positively.

In addition, participants had a generally positive attitude toward genetic tests, choosing to add more tests for newborns and committing to the outcome of premarital testing. These results are encouraging and indicate that there would be an overall positive response to introduce new genetic testing in the healthcare system. Furthermore, these findings, consistent with other reports, showed that Saudi society has a good attitude toward genetic testing, such as prenatal and preimplantation genetic diagnosis (14-16). Unexpectedly, most of the survey respondents had very little awareness of available health services. Most had never contacted a genetic counselor, and many were unaware of the presence or absence of genetic counseling facilities in their cities. Furthermore, many participants did not know where to access genetic tests for their families, and most considered health services for patients with genetic diseases to be rudimentary. This means that there is a lack of awareness and knowledge toward health services. This suggests that the launch of a national

Table S1. *Participant's awareness toward health service for genetic diseases.*

	Items	Responses	*N* (%)
1.	Do you know where you go to check whether your family carries a genetic disease or not and what type?	Yes	39.70%
		No	60.30%
2.	Do you know what you ought to do if you know that your children might be affected by genetic disease?	Yes	33.90%
		No	66.10%
3.	Do you agree to add more genetically related tests in pre-marital testing programs?	Yes	91.90%
		No	8.10%
4.	Have you ever got in touch with a genetic counsellor	Yes	3.90%
		No	96.10%
5.	Do you have genetic counselling facilities in your city/town?	Yes	15%
		No	20.80%
		Don't Know	64.20%
6.	Do you think that the health services provided for genetic diseases patients are advanced?	Yes	31.40%
		No	68.60%

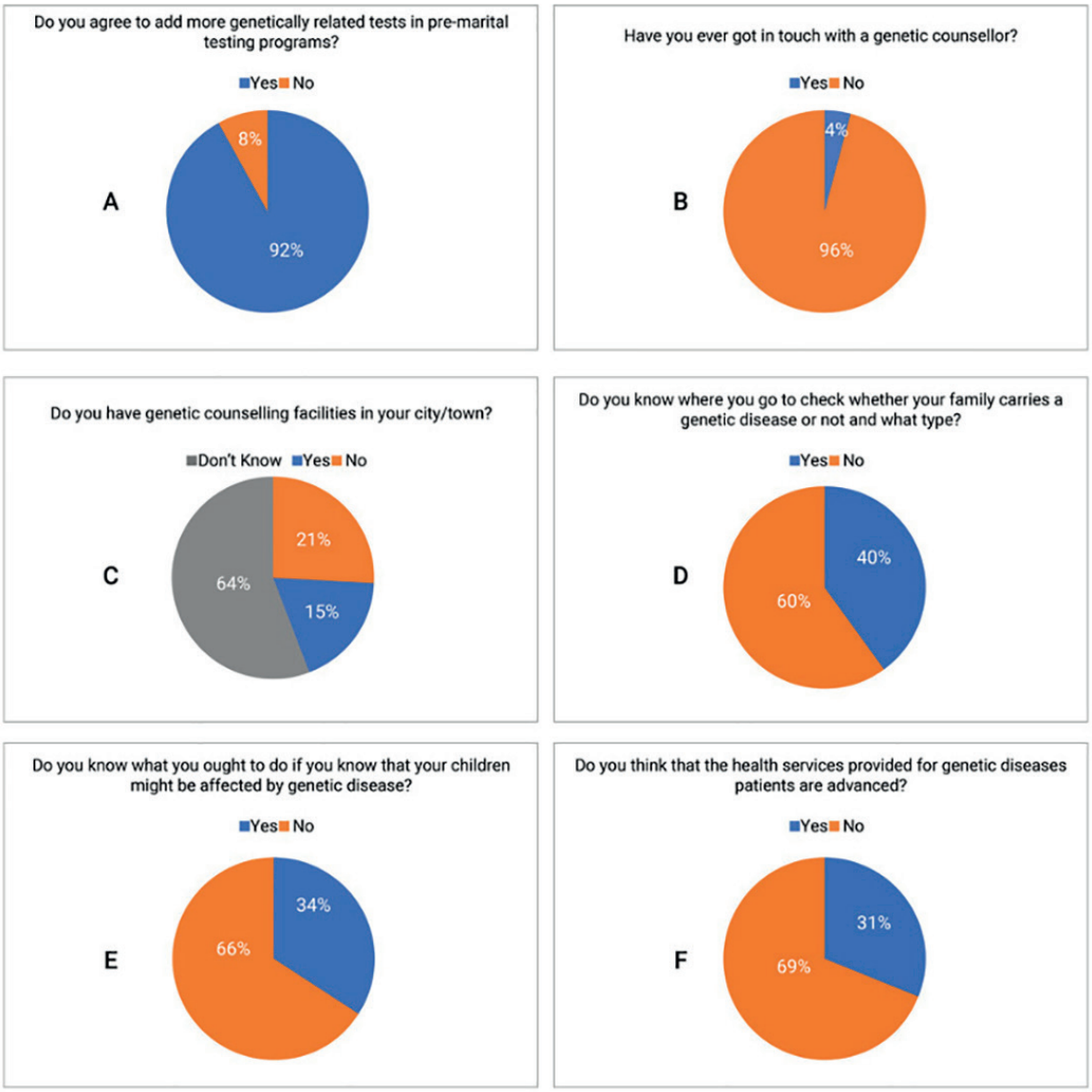

Figure 3. *Participant's awareness toward health service for genetic diseases.*

awareness campaign from health service providers explaining how to benefit from their services is essential.

To the best of authors' knowledge, few studies have evaluated the general understanding of genetic diseases in Saudi Arabia. The research is the first of its kind, and its findings will help policymakers who are dealing with genetic disease prevention in Saudi Arabia. The limitation of this study could be that it was conducted only in cities of the western region of Saudi Arabia. Therefore, we encourage health service providers to conduct surveys in all Saudi cities and launch campaigns about genetic disorders in schools, universities, shopping malls, and social media and websites. The increased awareness and knowledge regarding genetic diseases among the public will decrease the negative impact of genetic diseases in Saudi Arabia.

In addition, the Saudi human genome project (SHGP) will provide new opportunities to discover common and rare genetic disorders in Saudi Arabia (17). Thus, tests for these newly detected genetic diseases could be included premarital screening and newborn testing. Notably, the SHGP should plan for campaigns to improve the awareness and knowledge of genetic diseases among Saudi society.

Conclusion

In this study, we found that Saudi society had a high level of knowledge toward genetic diseases and had a good attitude toward genetic testing, such as prenatal and preimplantation genetic diagnosis. On other hand, Many of the study participants were not aware, where to access genetic tests for their families, and most considered health services for patients with genetic diseases to be rudimentary, thus pointing out a lack of awareness and knowledge toward health services. Therefore, awareness campaigns are needed.

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Author contribution

AFA, ABA, ISA, IYT, MA, RSA, and SHQ designed the preliminary survey draft. AFA and SHQ revised final survey. ABA, ISA, IYT, MA, and RSA distributed the survey through social media channels. AFA, ABA, ISA, IYT, MA, and RSA conducted analysis on the survey data. All authors wrote, reviewed, and approved the final manuscript.

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Declaration of conflicting interests

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

Consent to participate

Informed consent was obtained from the participants

Author details

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Supplementary Material (Table S1)

English version of the survey

Awareness and knowledge of genetic diseases among residents of the western region of Saudi Arabia

Section one (Personal Data)

Name: (optional)

Age: 10-15, 16-20 21-25. 26-35.36 and above

Gender: (optional)

- Educational level (secondary - university – diploma – post graduate - others)

Madinah (Mecca - Medina - Jeddah - Yanbu -)

Section Two

- In your opinion, How common is genetic diseases in the western region ?

(common - not widespread - I do not know)

- What are the most common genetic diseases in the western region?

(Cite three of them)

- In your opinion, what is the reason for the common of genetic diseases in the western region?

(Inbreeding - genetic mutations - lack of health awareness - others)

- Do you think that inbreeding is a cause of genetic diseases

(yes - no)

- Do you think genetic diseases are transmitted through infection?

(Yes - No)

- Is there anyone in your family born with a genetic disease ?

(yes - no)

“If the answer is yes, please specify the relationship of kinship and the disease”

- Did you received an awareness letter or benefit from a media campaign about genetic diseases?

(Yes – No)

Section three

- Do you know what are the most common genetic diseases in Saudi Arabia? (Yes - No)

“If yes, mention five of them”

- Do you know how you can reduce the risk of developing genetic diseases?

(Yes - No)

- Do you know where to go to find out if you have a genetic disease in your family?

(Yes No)

- Do you know what you should do if you know that you have the possibility of passing on a genetic disease to your children ?

(yes - no)

- Do you support the increase in the number of genetic diseases that must be tested in the pre-marital examination?

(yes - no)

- Do you know how genetic diseases are diagnosed ?

(yes - no)

- What is the best way to prevent genetic diseases?

(Raising health awareness - awareness campaigns in educational institutions - addition to testing for genetic diseases for newborns - commitment to the results of the prenuptial examination - other)

“You can choose more than one answer”

Section four

- Can people They were diagnosed with genetic diseases have healthy babies?

(Yes – No)

- Are the health services provided to people with genetic diseases developed? (Yes - No)

- Are there genetic counseling clinics in your city?

(Yes -No)

- Have you ever contacted a genetic counselor?

(Yes - No)

- Most of the time you get medical information about genetic diseases from:-

(Social networking sites - Internet pages - A specialist doctor - Other sources)

- Your level of awareness of genetic diseases and their seriousness:-

(High - Average – Low)