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Perception of Saudi Population in Al-Ahsa on Sickle Cell Disease and Sickle Cell Trait in Relation to The Genetic Screening Program

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ABSTRACT

Background: *Sickle cell disease takes an exceptional place among the most common inherited disorders in Saudi Arabia, remarkably in the Eastern Province of the Kingdom. Understanding the disease and its effects as a result of consanguineous marriages is critical. The level of perception among the Saudi population regarding genetic testing and screening needs to be measured. Therefore, the main objectives of this study were to assess the current health knowledge and beliefs of the Saudi population in Al-Ahsa regarding Sickle cell disease and Sickle cell trait, and how this population perceived genetic testing and screening program of sickle cell disease and sickle cell trait in order to establish baseline data.*

Methods: *This is a cross-sectional descriptive study of 205 Saudi subjects who attended the "Health Matters Seminar 2" (January 7-11, 2016). The subjects were selected by a purposive sampling technique, using a self-administered structured questionnaire.*

Results: *The study cohort comprised 205 Saudi subjects (32% male and 68% female) ranging in age between 10-60 years old. The subjects were interviewed and more than half (62.9%) had limited knowledge on how sickle cell disease and sickle cell trait are inherited.*

Conclusion: *There is a need to raise awareness on how sickle cell disease and sickle cell trait are inherited. In addition, there is a need to provide genetic counseling for the Saudi population in Al-Ahsa as part of the pre-marital screening (PMS) program.*

Keywords

Sickle Cell Disease, Health Beliefs, Perception, Attitude, Awareness, Knowledge, Genetic Screening, Genetic Testing and Counseling.

Introduction and Background

Sickle cell disease (SCD) is at the top of the most common hematological disorders hierarchy that causes serious and even severe consequences, which threaten an individual's life. It is caused by a particular mutation in the genetic makeup of the hemoglobin, a protein molecule that enables RBCs to perform their function, which is transporting of oxygen and carbon dioxide between lungs and body tissues. Based on this mutation, individuals with SCD suffer from long-lasting complications including severe painful episodes, anemia, stroke, infections, tissue damage, organ failure, and premature death [1].

SCD represents one of the severe hemolytic anemia forms [2]. SCD is caused by mutations in the hemoglobin beta genes. Typically, two normal beta globulin genes are inherited in normal individuals, one from each parent. SCD is an autosomal recessive genetic disorder, thus two defective beta-globulin genes (HbSs) are inherited, one from the mother and one from the father, in order for the disease state to occur. However, if an individual inherits only one defective hemoglobin (HbS) from one parent, that individual is diagnosed as a carrier for the disease and has sickle cell trait (SCT). Although having SCT means the individual is in good health and will not develop any symptoms of the disease, that individual has a 50%

chance of passing the trait to his/her children [3].

Severity and symptoms of sickle cell anemia depends mostly on the amount of Hb S. Sickled cells have a short lifespan, ranging from 10 to 12 days, due to the rapid blood hemolysis. Hemoglobin values usually range from 7 to 10 g/dL, thus anemia is present. Jaundice is one of the most obvious clinical signs too, and is usually reflected and seen in the sclera [4].

To offset the anemia, the bone marrow initiates the compensation mechanism by expanding, which leads to an enlargement of the face and skull bones. When the anemia becomes chronic, it can be associated with tachycardia, murmurs, and sometimes cardiomegaly, subsequently leading to dysrhythmias and heart failure in adulthood [3].

Facts about the prevalence of SCD in Saudi Arabia are limited and can be considered as underestimated, however, studies have reported that SCD belongs to the list of the most common inherited disorders whereas the first case was reported in the eastern region in 1960s [5]. In 2007, the highest rate of sickling was reported in Al-Ahsa region of the kingdom by Al-Hamdan and colleagues [6] (sickle cell disease 1.20%, and sickle cell trait 16.89%). Other regions of the kingdom showed much lower prevalence rates.

According to the Saudi research in 2008, the eastern region had the highest number of SCD cases with a prevalence of 145 cases out of 10,000 population compared with 24 cases out of 10,000 population in the southern region, 12 cases out of 10,000 population in the western region, and only 6 cases out of 10,000 population in the central region [7]. In 2011, Jastaniah [8] reported the variety of the prevalence of SCD in Saudi Arabia pointing out that the highest currency of the disease was reported in the Eastern province, then southwestern provinces to become after. Moreover, he reported that the currency of SCT ranged between 2% and 27%, and it reached to 2.6% of SCD cases in some regions.

This study aims to identify the perception of sickle cell disease among Saudi population in Al-Ahsa, as it is one of the regions that represent the high incidence of SCD, determine the current health knowledge of the Saudi public regarding SCD and SCT, and estimate the perception regarding the genetic screening among the Saudi population.

Research Methodology

This is a quantitative descriptive cross-sectional study. The study was conducted at "Your Health Matters Seminar 2". A seminar organized in Ibrahim Palace in Al-Ahsa region in Saudi Arabia on January 7-11, 2016. It involves 205 subjects. All of the subjects were contacted, counseled, and their verbal consents were obtained. Out of the 300 subjects approached, who visited our corner in the seminar, only 205 returned completed questionnaires, representing a response rate of 68.3%. Structured self-administrated questionnaires were distributed and collected on the same day. The purposive sample was made up of all the subjects who attended the seminar and were invited to answer the questionnaire anonymously.

Inclusion criteria

Initially, inclusion criteria were defined as: (1) Age ranges between 10 to 60 years; (2) Attendance at Your Health Matters Seminar 2; (3) Parent or guardian consent for those under 18 years of age. Applicants were excluded if they were non-Saudi.

Data Collection and Analysis

Subjects were asked to complete an anonymous questionnaire during the seminar, which was basically by the researchers. Approval for the study was obtained from the institutional Health Club Board in the university and confirmed by the Research Committee and the administrative board of the College of Applied Medical Sciences at King Faisal University. The subjects were assured that their participation would be voluntary. The purpose of the study was clarified to all subjects, and their consent was taken verbally before they were included in the study.

The questionnaire consisted of three sections to obtain the following information: The demographic data, knowledge and awareness regarding SCD, and common health beliefs of the individuals. In the questionnaire, a three-point rating scale was selected to assess the health awareness and beliefs of the subjects, with limiting the responses into three categories: agree, disagree, and uncertain. Researchers helped to administer the questionnaire to subjects who were illiterate and initiated to assist them while answering the questionnaire.

Data were analyzed using the Statistical Package for the Social Sciences (SPSS), Version 20. Socio-demographic variables considered in the analysis included sex, age, education level, marital status, and health status of family members. For each correct answer, a score of 1 was considered, then the total score was calculated to determine the level of knowledge regarding SCD. Subsequently, the total scores were classified into two categories. First category contains all scores of seven (7) and above; and evaluated as good knowledge. Second category contains all scores of six (6) and below which was then evaluated as limited knowledge.

Results

A total of 205 out of 300 Saudi individuals participated in the study (response rate: 68.3%). Of these, 65 were male (31.7%) and 140 were female (68.29%). Subjects ranged in age between 10-60 years old (mode: 21-30 years). Majority of the subjects (n = 81; 39.5 %) were between 21–30 years old.

Many of the subjects (n = 129; 62.9%) were married, and 76 (37 %) were unmarried. Most of the subjects (n = 94; 45.9%) were university students, and 71 (34.6 %) were hig school students. In relation to diagnosis with SCD, majority of the subjects were not patients (88.8%), and only (11.2%) were patients. On the other hand, (34%) reported that they have at least one of their family members diagnosed with SCD, whereas (66%) reported they have not at all.

Demographic data		No.	Percentage %
Age	10-20	42	20.6
	21-30	81	39.5
	31-40	54	26.4
	41-50	20	9.9
	51-60	8	3.5
Educational level	Illiterate or Primary	21	10.2
	High school	71	34.6
	University	94	45.9
	Post graduated study	19	9.3
Sex	Male	65	32
	Female	140	68
Patient	Yes	23	11.2
	No	182	88.8
Patients among family members	Yes	70	34
	No	135	66

Table 1: Distribution of Demographic Variable.

In terms of SCD knowledge and awareness, majority of the subjects (n=168, 82%) were correctly aware that sickle cell disease affects red blood cells, 109 subjects (53%)were also aware that SCD makes red blood cells hard and sickle shaped, 138 subjects (67.3%) knew that there are two genes causing SCD one from dad and one from mom, and 179 subjects (87.3%) agreed that we can know that someone carries the gene for SCD with a simple blood test.

Furthermore, many of the subjects (n= 126, 61.5 %) agreed that children with sickle cell disease are at risk for infections, pain, pneumonia and stroke. However, fewer subjects (n= 74, 36%) were aware that genetic carriers will not develop symptoms of sickle cell disease.

	Awareness and Beliefs	Agree (No.)	Disagree (No.)	Uncertain (No.)
1	Sickle cell disease affects the red blood cells	168	12	25
2	Children with sickle cell disease are at risk for infections, pain, pneumonia and stroke	126	35	44
3	Genetic carriers will not develop symptoms of sickle cell disease	74	119	12
4	Both parents must be carriers of sickle cell trait in order to have a baby with sickle cell disease	90	94	21
5	If only one parent is a carrier of a sickle cell trait, there is no chance of having a baby with sickle cell disease	87	87	31
6	I support sickle cell disease carrier testing	192	11	2
7	There are two genes causing SCD one from dad and one from mom	138	45	22
8	Having a sickle cell trait would make me less confident about forming a family	110	61	34
9	SCD makes red blood cells hard and sickle shaped	109	39	57
10	We can know that someone carries the gene for SCD with a simple blood test	179	13	13

Table 2: Knowledge and Beliefs of the Subjects.

On the other hand, near half of the subjects 87 (42.4%) agreed that if only one parent is a carrier of a sickle cell trait, there is no chance of having a baby with sickle cell disease. In addition, 90 subjects (44%) agreed and were correctly aware that both parents must be carriers of sickle cell trait in order to have a baby with sickle cell disease, and 94 subjects (45.9%) unfortunately disagreed and were unaware.

In terms of health beliefs,192 subjects (93.7%) supported sickle cell disease carrier testing, and 110 subjects (53.7%) believed that having a sickle cell trait would make them less confident about forming a family.

Responses to knowledge and beliefs questions "total score"

Table three showed that more than half of the subjects (62.9%) had limited knowledge toward SCD with a mean score of 5.9.

Items	No.	Percentage	Mean and std. Deviation
Good score "7 or more"	76	37.1	5.936585 ± 3.21
Poor score"6 or less"	129	62.9	

Table 3: The Total Score Out of 10 and its Interpretation.

Discussion

This study demonstrated that more than two-thirds of the subjects (67.3%) agreed on the fact that there are two genes causing SCD, one from dad and one from mom. However, this result shows a lower proportion compared to the result found by Al-Azri, Al-Belushi, Al-Mamari, Davidson, and Mathew [9] in a study took place in five health centers in Oman 2016, whereas the majority of the subjects (80.0%) were aware that SCD is an inherited genetic disorder. Moreover, this result also shows a lower knowledge level compared to the result of a study conducted on 2018 among a general population in Al-Ahsa by Al Ali et al. [10]., who summarized that (94.6%) of the respondents have heard about SCD, and (87.3%) knew that SCD is an inherited disorder.

The study of Al Ali et al., on the other side, showed that only (28.1%) knew that SCD can be associated with life-threatening infections, and only (16.3%) knew that stroke is one of the SCD complications. In contrast, more than half of the subjects of this study (61.5 %) agreed that children with sickle cell disease are at risk for infections, pain, pneumonia and stroke.

Another Nigerian study, conducted by Olakunle, Kenneth, Olakekan, and Adenike [11] among secondary school students in Abuja on 2013, showed a better knowledge whereas (80.0%) of the respondents were aware of SCD as an inherited disorder. On the other hand, (83.0%) of the respondents supported the fact that SCD affects the red blood cells, which is correspondent with what

was found via this study.

However, around (54%) knew that SCD can only be diagnosed via a blood test compared to the high number of the subjects involved in this study (87.3%) who knew that carrying the gene for SCD can be detected with a simple blood test.

The number of sufferers in this study was around three times higher (11.2%), compared to the study conducted on 2014 by Gamit et al. [12] who reported only (4%) sufferers. Furthermore, in this study, the subjects were aware of the sickle cell status of their family members whereas (34%) reported that they have at least one of their family members diagnosed with SCD. However, in the study conducted by Gamit et al, only (17%) of the participants knew the sickle cell status of their family members.

This study demonstrated that more than half of the subjects (62.9 %) had limited knowledge toward SCD. In contrast, a study carried by Abdulrahman Alghamdi et al. [13], to assess perceptions about SCD among the general population in Albaha region in Saudi Arabia, showed (68.80%) had good knowledge about basics of sickle cell disease.

Al-Farsi et al. [14], in a study of 400 Omani adults on 2014, reported that (84.5%) of the participants believed in the necessity of the premarital carrier screening. In consistence with this, (93.7%) of the subjects involved in this study supported sickle cell disease carrier testing.

Limitations

The small sample size, which was selected from a single area interviewed over a limited period, is one of the study limitations that affects our ability to generalize the findings. In order to validate the findings gained via this study, interviewing a larger sample size in future research is recommended. The recruitment criteria could also affect the study results because subjects filled the questionnaire at the sickle cell community campaign due to the lack of knowledge about sickle cell disease or due to their interest. The sampling method applied is prone to bias due to the lack of randomization.

Conclusion

This study reflects the current perception of Saudi population regarding SCD and SCT, and determines the level of acceptance of the genetic screening. Maintaining up-to-date research regarding SCD and its genetic issue in Saudi Arabia is critical for estimating the actual burden every year, and measuring the productiveness of the current strategies and approaches. Knowledge acquired by this study can help healthcare providers, nurses in particular, identifying the areas that need health promotion.

Vulnerability can be found among those who are affected by SCD; they are underserved and neglected although they experience a chronic disease that has an intense influence on their lives. SCD population needs education, health promotion and awareness programs so they can appreciate their health and the importance of PMS program. The knowledge acquired by this study can contribute to improve community awareness regarding the importance of understanding the disease and appreciating the genetic testing required.

Genetic counseling is a necessary discipline as it relates to the future of a community's health. Saudi Arabia, as one of the developing countries that have a high prevalence of SCD, must increase awareness in communities, schools, medical associations, health institutions and through the media; and must stand beside all SCD patients by incorporating support and advocacy.

Although the genetic counseling and PMS program have been successful, around half of the subjects in this study (53.7%) were less confident about forming a family. Therefore, Saudi Arabia must implement other strategies that can be able to identify the cases at early stage, through newborn screening for instance, and must focus on the strategies that can reduce SCD-related morbidly and mortality such as applying parental counseling and providing a continuation of care for the patients. Finally, Saudi Arabia must plan for more advanced and early awareness programs to make people more confident and able to overcome any barriers.

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