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PREMARITAL GENETIC TESTING IN PRIMARY CARE CENTERS IN SAUDI ARABIA; A SYSTEMATIC REVIEW

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Abstract:

Background: *Genetic abnormalities in the Gulf area are increasing especially heamoglobinopathy and sickle cell disease. This can be attributed to increased rate of consanguineous marriage. Therefore, premarital genetic testing can play an important role in minimizing genetic abnormalities in the community.****Objective:*** *The aim of this systematic review is to evaluate the literature to describe premarital genetic screening in primary care units particularly in Saudi Arabia.****Method:*** *the literature was reviewed through PubMed database in the duration between 2008 and 2018. Search terms included were a combination of '' premarital, 'genetic screening and ''Saudi Arabia''. The results were then filtered to include original research articles investigating premarital genetic testing in primary care units in Saudi Arabia. Selected trials mentioned the targeted age group in addition to the genetic abnormality targeted by the screening program.****Result:*** *A total of 72 articles were retrieved. Following including only trials in Saudi Arabia, 23 articles appeared. After checking for the eligibility criteria, a total of six articles were considered as eligible to be included in our systematic review that were published between 2008 and 2018 covering a total of 1,576,496 Saudi citizens. Four trials included young adult subjects. While the other two trials included subjects with a wider range of age, from 19 to 50 and from 17 to 58 years old, respectively. All the trials examined the importance of genetic testing for both thalassemia and sickle cell anemia either through assessing knowledge and awareness of the community toward the importance of this test or through determining the rate of at risk marriages; however, Olwi et al examined thalassemia only.* ***Conclusion:*** *Awareness towards genetic testing needs to be increased in the Saudi Arabian community. More genetic abnormalities need to be included in premarital genetic screening.*

Keywords: *Premarital, genetic testing, congenital diseases, sickle cell anemia, thalassemia.*

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INTRODUCTION:

A wide range of Arab nations demonstrate a high level of genetic variety. Main contributors that caused such diversity comprise the immigrations from other parts of the world to Arab countries [1]. In many situations these immigrations led to important mixes of Arab people with other nations from Africa, Europe, East and South Asia [2].

Trials evaluating the incidence of genetic disorders in addition to congenital abnormalities that are fatal or lead to long term disability if not managed showed that, the highest incidence of affected children was recorded from the Arab countries compared to western countries [3].

Moreover, there is an increasing prevalence of genetic abnormalities like hemoglobinopathies in Middle East region, with a rate of carriers ranging from 2.5 to 27% for G6PD deficiency, and about 50% for thalassemia, also incidence of sickle cell anemia was shown to be almost 30% in Arab countries [4].

In parallel to the big number of members in many Arab families, there are high consanguinity rates that increased the possibility of occurrence of various autosomal recessive abnormalities [5], in particular rare or previously unreported syndromes and metabolic defects [6].

Consanguineous marriage remains to be adopted in many Arab countries, with similar disease phenotype frequencies to those recorded in their original countries [7], comprising a large fraction of autosomal recessive cases [8].

Premarital genetic screening strategies for thalassemia in addition to other hemoglobinopathies are currently mandatory in a many Middle Eastern country [9], where couple is referred to genetic counseling clinic if both are found to be carriers [10]. Therefore, the aim of this systematic review is to examine the literature to describe the figures of premarital genetic screening in primary care units in Saudi Arabia.

METHODOLOGY:

This systemic review of literature was done on PubMed database in the duration between 2008 and 2018, in order to evaluate premarital genetic testing in primary care units in Saudi Arabia. Search terms included were a combination of ‘’ premarital, ‘genetic screening and ‘‘Saudi Arabia’’.

All the titles in addition to abstracts appeared from this search were reviewed thoroughly. The results were then filtered to include original research articles investigating premarital genetic testing in primary care units in Saudi Arabia. Additionally, the selected trials specified the screened genetic abnormality only performed on premarital basis. Only trials published in English language were classified as related articles which can be further evaluated in the second step.

The following step was determining the inclusion criteria to choose the studies that will be considered in the systematic review. Abstracts were revised manually to choose the appropriate abstracts to be considered. The inclusion criteria were the presence of sufficient details on the screened genetic abnormality and the targeted age group. Moreover, only trials done in Saudi Arabia were included.

Furthermore, references of selected trials were revised in order to define any related articles. Finally, the required data sets were collected from the final record of eligible articles and summarized.

Data was statistically described in terms of frequencies (number of cases) and valid percentages for categorical variables. Mean, standard deviations, minimum and maximum were used to describe numerical variable. All statistical calculations were done using computer program IBM SPSS (Statistical Package for the Social Science; IBM Corp, Armonk, NY, USA) release 21 for Microsoft Windows.

Before conducting any study related procedures, institutional approval was obtained. There was no need to obtain consent form as the study is not involving any interventions on patients.

RESULTS:

A total of 72 articles were retrieved by searching *PubMed* using the combination of the three terms ‘’ premarital, ‘genetic screening and ‘‘Saudi Arabia’’. Following exclusion of articles in all parts in the world and including only trials in Saudi Arabia, 23 articles appeared.

After searching the abstracts and checking for the eligibility criteria in identified potential abstracts, a total of six articles [11-16] were considered as eligible to be included in our systematic review that were published between 2008 and 2018 covering a total of 1,576,496 Saudi citizens.

Table 1. Summary of included studied and their results

Author(s)	Year	Study design	Sample size	Targeted age group	Screened genetic diseases	Objective	Result
Olwi *et al* [11]	2017	A cross-sectional survey	920	mean age 22.4 ± 1.3 years	Thalassemia	assess college students' knowledge of importance of genetic testing to predict thalassemia.	A substantial proportion of the participants had a low knowledge of thalassemia. This lack of awareness requires a reassessment of the goals and success of the premarital testing program, including the genetic counseling services, and also indicates the importance of emphasizing thalassemia in school curricula and promoting and scaling up existing thalassemia campaigns in the region.
Ibrahim *et al* [12]	2012	cross-sectional study	655	Mean age of participants was 30.63 ± 9.1 years.	Sickle cell anemia and thalassemia.	assess the knowledge and attitude of individuals attending governmental outpatient clinics regarding the Premarital Screening and Genetic Counseling (PMSGC) programs, to identify predictors of high knowledge scores and to determine the satisfaction and recommendations of clients of the program.	Knowledge in the general population about the PMSGC program was low. Implementation of school and university educational campaigns is important. Improved counseling and adding new topics for counseling on genetic, chronic, and psychiatric problems; building healthy families; reproduction and fertility are recommended.
Alswaidi *et al* [13]	2012	case–controlled study	934	The age ranges of 19-39 years old.	sickle cell disease and β-thalassemia	determined the rate of at-risk marriages and identified several factors that may prevent at risk couples from marrying due to possibility of	Most couples received no advice to participate in genetic counseling services. Marriage decisions for the small number who received genetic counseling did not differ significantly from those who received no counseling.

						genetic disorder in off springs. in order to assess relationships between various cultural and social factors and marriage decisions.	Recommendations are made for improving the effectiveness of this screening program.
Memish *et al* [14]	2011	Retrospective	1572140	19-50	sickle cell disease and b-thalassemia	Regional differences in the prevalence of sickle cell disease and thalassemia, as well as the frequency of at-risk marriages using 6 years of national data from the premarital screening &genetic counseling program.	Vast regional differences in hemoglobinopathies among adult Saudis are being reported that may help policy makers better allocate resources of available preventive programs.
Al-Aama *et al* [15]	2010	cross-sectional study	800	aged 18–29	sickle cell disease and b-thalassemia	To examine the attitude of young educated individuals regarding the national Premarital genetic screening program and its implementation.	Further public education and wide spread genetic counseling prior to testing is essential for the success of the program. Screening singles on admission to university prior to any commitment may be preferable than screening immediately before the marriage certificate is issued.
Al Sulaiman *et al* [16]	2008	cross-sectional survey study	1047	median age 28.1, range 17–58 years.	control inherited hemoglobin (Hb) disorders	to assess the knowledge, perception, and attitude among the Saudi population about the premarital genetic screening program through a questionnaire-based survey.	The concept of genetic counseling was liked by most of the participants. There was a positive attitude toward the program and the majority agreed to apply the PMS program to all couple in all country regions. More than 60% of all the participants were in favor of preventing at-risk marriages.

Out of the total number of citizens, four trials [11-13, 15] included young adult subjects with age ranging between 20 and 30. On the other hand, Memish *et al* [14] and Al Sulaiman *et al* [16] included subjects with a wider range of age, from 19 to 50 and from 17 to 58 years old, respectively.

Turning to study design, all the studies had a cross sectional design except for Memish *et al* [14] had a retrospective design. Additionally, Alswaidi *et al* [13] was the only case control study included in this review.

According to extracted results, all the trials examined the importance of genetic testing for both thalassemia and sickle cell anemia either through assessing knowledge and awareness of the community toward the importance of this test or through determining the rate of at risk marriages, however, Olwi *et al* [11] examined thalassemia only . The included trials are discussed in details in table 1.

DISCUSSION:

Incidence of genetic and hereditary diseases is increasing drastically during the past decades especially in Gulf area. That's why there is a demanding need to highlight the importance of genetic screening and counseling services before marriage in order to decrease the rate of these abnormalities that represents a burden on the community from social and economic perspectives.

The present work evaluated the medical literature through PubMed database to describe the figures for premarital genetic screening and counseling in primary care units in Saudi Arabia. Six studies [11-16] described the situation in Saudi Arabia either in terms of the knowledge and awareness of the community about premarital genetic testing or through investigating the incidence and consequences of at risk marriages.

From the present systematic review, it was found that the level of knowledge and awareness in the Saudi community toward genetic diseases, mainly thalassemia and sickle cell disease, is considered unsatisfactory and needs to be improved. Additionally, the knowledge and awareness of the community towards genetic screening and premarital counseling is considered low as well. Furthermore, most of the couples preparing for marriage don't get an advice to go for a genetic counseling service before marriage.

Most of the included studies aimed at evaluating the knowledge and awareness of the general population in Saudi Arabia. Only Olwi *et al* [11] focused mainly on students' awareness and knowledge. This is of great significance because increased awareness at younger age can prevent at risk marriage at any earlier stage rather than immediately before signing marriage contract.

Olwi *et al* [11] concluded that the level of knowledge of the students was considered low and needs more awareness programs to improve their level of knowledge. This conclusion was compliant to the findings of other trials in different age groups. For instance, Ibrahim *et al* [12], Al Aama *et al* [15] and Al Sulaiman *et al* [16] reported a low level of knowledge and awareness of the Saudi population on premarital genetic testing and counseling services.

It is worth to mention that Olwi *et al* [11] targeted only thalassemia in their study, while Ibrahim *et al* [12], Al Aama et al[15] focused on both Sickle cell anemia and thalassemia, the most two prevalent genetic disorders in Saudi Arabia. Finally, Al Sulaiman *et al* [16] were concerned by inherited hemoglobin disorders without specifying which one of them.

Not only knowledge and awareness towards genetic testing and counseling was evaluated, the prevalence and distribution of genetic disorders all over the Kingdom was also examined through the mega trial of Memish *et al* [14]. This study included records of 1,572,140 Saudi citizens in order to describe the figures of prevalence of sickle cell anemia and thalassemia in addition to the rate of at risk marriages in Saudi Arabia.

Memish *et al* [14] showed that there is a wide variation in the prevalence of genetic disorders among different areas in Saudi Arabia which can help decision makers to focus more on areas with highest prevalence. In spite of the large sample size of this study, it had a retrospective design.

Finally, all the six trials [11-16] strongly recommended increasing educational and awareness programs all over Saudi Arabia in order to increase the knowledge of the population on genetic disorders and the importance of responding to the premarital genetic screening and counseling in order to minimize the incidence of this problem in the Saudi community.

To our knowledge, this is the first updated systematic review that shows the figures for premarital genetic testing in primary care units in Saudi Arabia during the last decade.

CONCLUSION:

The burden of genetic disorders in Saudi Arabia is alarming and needs rapid and effective actions especially with the low levels of awareness and knowledge of the Saudi citizens on the prevalent genetic disorders in Saudi Arabia and genetic screening and counseling. Educational programs and awareness campaign should target a large sector of the community of different age groups and levels of education in order to increase the knowledge and awareness of the community.

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