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Research Article

FREQUENCY OF HAEMOGLOBINOPATHIES AMONG ATTENDANTS OF CENTRAL LABORATORY AND BLOOD BANK AL-BAHA KSA

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ABSTRACT

Background: Haemoglobinopathies are the most frequent single gene disorders worldwide, and particularly in the Eastern Mediterranean region, including Saudi Arabia. Information about the prevalence of sickle cell disease and thalassemia in the Southern region of Saudi including Al-Baha is not clear. The aim of this study was an attempt to determine the frequency of haemoglobinopathies among the individuals screened as part of the Saudi Premarital Screening Program at the Central Laboratory and Blood Bank Al-Baha, Saudi Arabia

Methods: this was a retrospective cross-sectional study was conducted at the Central Laboratory and Blood Bank Al-Baha, Saudi Arabia as part of the National Premarital Screening Program. Reports of the years 1435 and 1436 Hijra (February 2014 to January 2016) were reviewed.

Results: Totally 4,055 individuals were screened, the detected 278 cases with abnormal hemoglobin constitute (6.83%) of the screened samples during the period of the study ,with sickle cell trait (3.7%), followed by β -thalassemia trait, sickle cell disease with (1.3 %) and (0.99 %) respectively , while variant haemoglobin constitute only (0.86%)

Conclusion: We conclude that sickle cell anemia (SCA) is the most common type of haemoglobinopathies among the population of Al-Baha region, and the screening program constitute the backbone of the preventive strategy for these haemoglobinopathies.

Recommendation: Family screening of the SCA and SCT is necessary to identify sickle cell carriers that should be extend to all areas of Al-Baha region

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INTRODUCTION

The haemoglobinopathies are genetic diseases caused by a range of different deletional and non-deletional mutations of globin genes, which determine the structure as well as the amount of the various globin chains of the hemoglobin molecule.¹ They are the most frequent single gene disorders worldwide, and particularly in the Eastern Mediterranean region, including Saudi Arabia², which has a high prevalence of hereditary hemoglobin disorders.³ The haemoglobinopathies are categorized into two main groups: - the hemoglobin variants and the thalassemia. The hemoglobin variants arise from an alteration in the globin protein structure whereas the thalassemia arise from inadequate production of the normal globin protein. Sickle cell disorders often result in severe life threatening clinical symptoms. Those patients with B thalassemia major require regular blood transfusions to

maintain life. According to the WHO, approximately 240 million people are heterozygous for inherited hemoglobinopathies, including thalassemia and sickle cell disease.⁴ Premarital screening programs for thalassemia and sickle cell disorders aim to identify asymptomatic carriers of these autosomal recessive disorders, so that they are informed and can understand their reproductive risks and available options.⁵ WHO described pre-marital genetic screening as services targeted at individuals and families which try to enable people with genetic disadvantages, and their families to live and reproduce as normally as possible, assuring access to relevant medical services and social support systems, helping them to adapt to their unique situation and providing information to enable educated and voluntary choices in health and reproductive matters.⁶

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Saudi Arabia has a mandatory premarital screening program called Healthy Marriage where screening includes only haemoglobinopathies, Human Immunodeficiency Virus (HIV), Hepatitis B Virus (HBV), and Hepatitis C Virus (HCV).⁷ Information about the prevalence of sickle cell disease and thalassemia in the Southern region of Saudi including Al-Baha is not clear. A number of studies conducted in Saudi Arabia demonstrated that the prevalence of these diseases varied significantly in different parts of the country, with the highest prevalence in the Eastern province of the country, followed by southwestern provinces.^{8, 9} The reported prevalence for sickle-cell trait ranges from 2% to 27%, and up to 2.6% will have SCD in some areas¹⁰, in a study published in 2007 reporting 3.4% of 488,315 premarital screenings done have a thalassemia trait, while 0.07% had the thalassemia disease; focused mostly in the western, southwestern, and eastern parts of Saudi Arabia.¹¹

The aim of this study was an attempt to determine the frequency of haemoglobinopathies among the individuals screened as part of the Saudi Premarital Screening Program at the Central Laboratory and Blood Bank Al-Baha, Saudi Arabia.

MATERIAL AND METHODS

Study design and setting: This is a descriptive cross-sectional study, for years 1435 H (2014) and 1436 H (2015), among individuals attending Central Laboratory and Blood Bank Al-Baha, who intended to get married, underwent routine mandatory tests and those with a complaint of anemia. This center is one of the accredited premarital screening centers throughout the Kingdom's regions and provinces in Al-Baha conducting examination for soon-to-be married in order to identify if there is any injury with genetic blood diseases such as sickle-cell anemia (SCA) and Thalassemia, and some infectious diseases such as hepatitis B, C and HIV "Aids", and thus, it receives individuals from all of Al- Baba region.

Data collection: A checklist was used to gain information for each individual of the those attending the medical laboratory during the study period were included. A sample of 5 mL blood in two different collecting tubes were taken from each individual by venipuncture, 3 ml in plain tube for viral testing and the other 2 ml in ethylenediamine tetra-acetic acid (EDTA) containing tube for complete blood count (CBC) and hemoglobin electrophoresis (HE). Hematological and red cell indices were estimated by an automated cell counter, and the sample was then used to perform the sickling test, if the mean corpuscular volume (MCV) was less than 80 fL and/or the mean corpuscular haemoglobin (MCH) less than 27 pg and/or the sickling test was positive , the sample was then processed further for the quantitation of Hb A2, Hb F, Hb S or any other variants, then haemoglobin typing was performed quantitatively by an automated ion exchange high performance liquid chromatography (HPLC) system on the Bio-Rad variant II instrument (Bio-Rad Laboratories, Belgium). β-thalassaemia trait was identified by the characteristic elevation of Hb A₂ (>3.8%). Hb S, Hb C, Hb D and others were detected according to their specific peak area that was calculated after elution with the buffer solution.

Data analysis: All statistical analysis was performed by using Excel sheets for frequencies and tabulations.

Ethical issues: Ethical clearance was given by the Research Committee in Al Baha University Faculty of Medicine and the health authorities concerned.

Conflict of interest: No potential conflict of interest was reported by the authors."

RESULTS

According to the annual reports for the years 1435 H (2014) and 1436 H (2015) reviewed, 4055 samples were processed during the study period, with male constituting 43.6 % (1768/4055) and female 56.4% (2287/4055) (Fig 1). The detected 278 cases with abnormal hemoglobin constitute (6.83%) of the screened samples during the period of the study. 56.2 % (156 / 278) were females & 43.8 % (122 /278) were males. The most predominant was sickle cell trait (3.7%) with females constituting 75%, followed by β-thalassemia trait, sickle cell disease with (1.3 %) and (0.99%) respectively, while variant haemoglobin constitute only (0.86%), as shown in (Fig.2)

Table 1 The frequencies of different haemoglobinopathies

	Male	Female	Total	%
No of Screened / year	1768	2287	4055	
Abnormal hemoglobin	122	156	278	6.83 %
Sickle cell trait	37	112	149	3.7 %
Sickle cell disease	15	25	40	0.99 %
β-thalassemia trait	24	30	54	1.3%
Variant haemoglobinopathies	15	20	35	0.86%

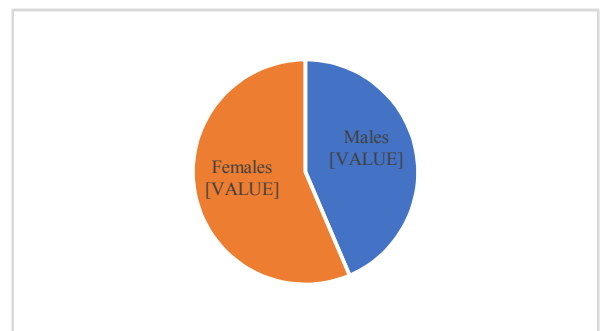


Fig1 Sex of the attendants

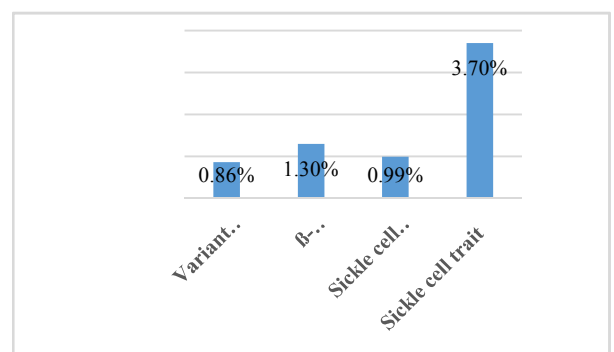


Fig 2 Frequencies Haemoglobinopathies among the attendants

DISCUSSION

Haemoglobinopathies are one of the major health problems in Saudi Arabia, especially in Southern, Western and Eastern areas where the gene frequency of this disease is quite prevalent. Many studies were carried to identify the prevalence and frequencies of haemoglobinopathies in these areas. To our

knowledge, studies concerning frequency of haemoglobinopathies were scarce in the area of Al-Baha.

In our study, we reviewed the reports of the *National Premarital Screening Program during the years 1435 H and 1436 H (February 2014 to January 2016)* mostly for premarital couples at the Central Laboratory and Blood Bank Al-Baha, Saudi Arabia, to determine the frequency of hemoglobinopathies in Al-Baha region. We have detected 278 cases with hemoglobinopathies. The overall frequency of haemoglobinopathies in our study was 6.83%; and the prevalence rate of sickle cell trait and sick cell disease, observed in this study were (3.7% and 0.99 % respectively) were comparable with those reported from Ziad A. Memish *et.al* (2011) among 43,208 men and women examined in Al-Baha centers of premarital screening with 3.6 % for sickle cell trait and 0.02 % for sickle cell disease,¹² again this findings were lower than findings of a study done by Al Jaouni SK *et.al* (2010) "Prevalence of thalassemia disorders and hemoglobinopathies in Jeddah, western Saudi Arabia" which showed the prevalence of sickle cell trait was 5.4%.¹³ The frequency of thalassemia trait (carriers) among individuals having haemoglobinopathies in our study was 1.3 % , compared with 0.1 % in study by Ziad A. Memish *et.al* (2011). The results of our study support the finding that sickle cell trait is the most frequent form of haemoglobinopathy in Al-Baha region, which coincide with findings of Ziad A. Memish *et.al* (2011).

This high frequency of haemoglobinopathies in the region reflects the high regional and geographical prevalence; this is partly because of the high ratio of consanguineous cousin marriages among Al-Baha resident population.

CONCLUSIONS

We conclude that sickle cell anemia (SCA) is the most common type of haemoglobinopathies among the population of Al-Baha region, and the screening program constitute the backbone of the preventive strategy for these haemoglobinopathies.

Recommendations

We recommend that family screening of the SCA and SCT is necessary to identify sickle cell carriers that should be extend to all areas of Al-Baha region, and that premarital screening program should be considered as a routine investigation, which should be strongly supported by the authorities.

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