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ORIGINAL ARTICLE

Hemoglobin Disorders Among Anemic Patients: a Cross-Sectional Study from Jeddah City, Western Saudi Arabia

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SUMMARY

Background: **Hemoglobinopathies and thalassemia are defined as a group of inherited blood disorders characterized by a variable degree of anemia with a wide spectrum of clinical symptoms. They are commonly found in the Mediterranean area, sub-Sahara Africa, Middle East, Central India, and Southeast Asia with an estimation of 400,000 babies born annually with serious hemoglobinopathies. Of those, 90% of the births occur in underdeveloped or developing countries. This study was undertaken to investigate the prevalence of hemoglobin disorders among anemic patients who visited a tertiary care setting represented by King Abdulaziz University Hospital.**
Methods: **This is a cross sectional study which investigated blood samples from 668 anemic patients for possible causes of anemia. This investigation involved the use of complete blood count, hemoglobin separation using capillary electrophoresis, and measurement of nutritional elements commonly investigated for anemia.**
Results: **We found that the frequency of different types of hemoglobinopathies and thalassemia among the subjects were as follow; normal (HbAA) 439 (65.7%); Sickle Cell Trait (HbAS) 65 (9.7%); Sickle Cell Anemia (HbSS) 63 (9.4%); β-thalassemia trait 48 (7.2%); Hb S/β 27 (4.0%); HbH 7 (1.0%); HbE 6 (0.9%); beta-thalassemia major 6 (0.9%); Hb E/beta-thalassemia 4 (0.6%); HbC 1 (0.1%); HbD 1 (0.1%) and HbSC 1 (0.1%).**
Conclusions: **The findings of this study emphasize the necessity of increasing public health education, neonatal and adult screening programs, as well as nutritional guidance and plans to start the eradication of this burden.**
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KEY WORDS

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INTRODUCTION

Hemoglobinopathies represent a major group of inherited monogenic disorders affecting 7% of the world population [1]. In Saudi Arabia, the prevalence of hemoglobinopathies varies from one region to another and consanguineous marriage is customary in some regions rendering it as the main factor for spreading such disorders [2]. It has been estimated that the prevalence of hemoglobinopathies in the Saudi population accounts for about 4.55% comprising mainly Beta-thalassemia and Sickle Cell Disorders [3]. There are several reasons for

the high frequency of hemoglobin disorders in many tropical countries. Basically, a theory of natural selection has been raised as among the causes through the protection feature of heterozygous state against malaria that is commonly found in the tropical countries [4]. Other factors include consanguineous marriages in many of these countries resulting in a wide inheritance of such genetic disorders. However, exact information on the occurrence rate of consanguineous marriage is not clear. It is without doubt, that such a factor maintains the burden of hemoglobinopathies on the health system. Another contributing factor is the epidemiological transition, with the improvement of public health of the population as well as adequate nourishment in the poorer countries, it is found that children who would have been died, enjoy many years of life with accurate diagnosis and proper management. This is typically seen in Cyprus. After the second world war it passed through this transitional stage [5]. At last, the different distribution of the hemoglobin diseases in various populations reflects strong founder impact by their original inhabitants which can be seen in the Pacific Island populations.

In the western province of Saudi Arabia, around 11.2% of its healthy population are affected with haemoglobinopathies. Among all hemoglobinopathies, sickle cell anemia represents the major form with a prevalence of 5.4% followed by beta (β)-thalassemia trait (4.69%), hemoglobin E (HbE) trait (0.85%), and HbH disease (0.19%) [6]. Presence of such disorders remains one of the major challenges encountered by health authorities since there is no complete cure despite the advancement of molecular biology tools and treatment strategies. Anemia, reflected by low hemoglobin level, indicates the need for hospitalization and probably blood transfusion, which may cause further complications to these patients. Investigating the prevalence of hemoglobin disorders among anemic patients who visit tertiary hospitals in this region of Saudi Arabia should be of widespread interest to healthcare professionals such as clinicians, genetic counselors, and molecular geneticists to know the status of hemoglobin disorders and their future burden on health resources. Therefore, it is important to identify the extent of hemoglobinopathies among anemic patients in order to be able to have a strategic health plan. This study aimed to investigate the prevalence of hemoglobin disorders among anemic patients who visited King Abdulaziz University Hospital in the western region of Saudi Arabia.

MATERIALS AND METHODS

Study subjects

A total of 668 individuals were recruited in this study. Inclusion criteria for the samples were to be from anemic patients regardless of age and gender. Ethical approval was obtained from the unit of biomedical ethics, Faculty of Medicine, KAU, with the number (78575/38/D - 2/7/1438 AH).

Blood samples and hematological analyses

Blood samples were collected from subjects at steady state using disposable vacutainer system in ethylenediaminetetraacetic acid (EDTA) tubes as an anticoagulant for hematological parameters and plain tubes for chemistry analysis. Complete Blood Count profiles were carried out on EDTA (Beckton and Dickinson) tubes using a Sysmex XE-2100 analyzer (Sysmex Corporation, Kobe, Japan). Where indicated, blood smears and films were done using Wright Giemsa stain (Merck KGaA, Darmstadt, Germany).

Sickle cell screening test

All samples were screened for the presence of hemoglobin S (HbS). The test is based on solubility principle where red blood cells (RBCs) are lysed by saponin reagent and the free hemoglobin is reduced by sodium hydrosulfite in a concentrated phosphate buffer. Presence of HbS can be detected through formation of crystals resulting in inability of solubility of HbS. Therefore, a turbid solution can be formed indicating a positive reaction. The negative reaction is reflected by a clear transparent solution upon lysis of RBCs indicating the absence of HbS. In the case that α-thalassemia intermedia is suspected, HbH inclusion bodies test was done using brilliant cresyl blue stain (BDH Chemicals, Middlesex, England).

Hemoglobin separation

Hemoglobin electrophoresis was carried out using Capillarys 2 flex piercing analyzer for electrophoresis. Its main principle depends on electroosmotic flow and pH, where the charged particles are separated via their mobility in an alkaline buffer (Sebia, Issy-les-Moulineaux, France).

Measurements of chemical elements

Iron profile was tested for samples with microcytic hypochromic anemia indicated by the complete blood count. This was performed to exclude iron deficiency anemia from samples suspected with thalassemia. The iron profile includes serum iron and ferritin. The test was performed on a Dimension Vista 1500 (Siemens Healthcare Diagnostics, Germany) machine. Serum iron was measured using a colorimetric method, while the measurement of ferritin was done using the chemiluminescent, sandwich technique.

Statistical analysis

Statistical analysis was performed using Excel in order to determine the frequency of hemoglobinopathies among anemic patients.

RESULTS

This study involved 668 patients with anemia who visited KAUH and were included in the study irrespective of their age and gender. Hemoglobinopathies including thalassemia are contemplated to be major causes of anemia in our community [7]. Therefore, in this study, we reviewed the burden they pose in anemic patients. Anemia can be divided into three categories based on its extent of intensity; severe (< 7.0 g/dL), moderate (7.1 - 10.0 g/dL), and mild (10.1 - 12.0 g/dL) [8]. Our study showed that most of our subjects have moderate anemia with a prevalence of 45.7%. However, the severe group, which is considered as critical and requires immediate intervention i.e., blood transfusion, constitutes 24.8% of the subjects. Table 1 shows the prevalence of mild anemia is 29.5% with a mean of Hb of 11.0 g/dL.

Anemia can additionally be classified with respect to morphological features of RBCs as seen on blood smears with reporting by automated cell counter indices [9]. Table 2 shows the distribution of anemia in the studied 668 subjects according to the mean corpuscular volume (MCV) and the mean corpuscular hemoglobin (MCH) results. The most prevalent type of Anemia in our study was noted to be microcytic hypochromic anemia with a prevalence of 79% (n = 528), as compared to a prevalence of 18% (n = 120) normocytic hypochromic type of anemia, and 3% (n = 17) with macrocytic anemia. Almost 98% (n = 654) of all anemic patients were hypochromic (low MCH). Since iron is considered an important element for RBC formation, Table 3 shows the iron profile parameters in subjects with the microcytic hypochromic subtype of anemia. The highest number (25.9%) (n = 137) of subjects were present in the low iron and low ferritin subgroup, while the lowest (1.7%) (n = 9) number of subjects were noted to be in the high iron and low ferritin subgroup. Normal iron and ferritin levels were found in (24.4%) (n = 129) of microcytic hypochromic patients Additionally, our results revealed that 23.4% (n = 156) of the recruited patients with anemia are positive for the sickle cell trait as shown in Table 4.

Table 5 shows how the various means of hematological parameters change with 8 abnormal variants of Hb, compared to the normal (HbAA). As shown, the highest number of RBC count was noted in β-thalassemia trait followed by HbE, HbH disease, and sickle cell trait with all these variants having more RBCs than in anemic patients with HbAA, while the lowest RBC numbers were seen in sickle cell disease, β-thalassemia major, HbE/β-thalassemia, HbS/β-thalassemia. Hb, hematocrit, and MCV were lowest in HbE/β-thalassemia disease among all variants of Hb and HbAA, while the highest Hb and hematocrit was found in HbE disease alone. However, the highest MCV was noted in sickle cell disease. RDW reflects the variability in the size of RBCs and is typically lowest in patients with normal HbAA, as seen in our study. The highest RDW was noted in HbE/β-thalassemia, followed by β-thalassemia alone. The reticulocyte count is a measure of the production of the bone marrow of new RBCs and ranges 0.5 - 1.5% in a normal individual. The highest number of reticulocytes were found in sickle cell disease followed by HbS/β-thalassemia combination and the lowest number of reticulocytes was found in β-thalassemia trait and major along with anemia with HbAA.

Table 6 shows the prevalence of different haemoglobinopathies in our 668 patients. Sickle cell disorder was found to be the most prevalent hemoglobinopathy causing anemia in our study with a frequency 9.73% for sickle cell trait, 9.43% for sickle cell anemia (HbSS), 4.04% for compound sickle/beta thalassemia and 0.15% for compound hemoglobin S/C disease comprising a total estimation of 23.35% collectively. This is followed by beta-thalassemia trait (7.19%) and HbH (1.0%). Minor frequencies of hemoglobinopathies with less than 1% were found to be beta thalassemia major and hemoglobin E disease comprising 0.9% per each. Compound hemoglobin E/beta thalassemia was also detected in our study with a prevalence of (0.6%) followed by hemoglobin C and hemoglobin D diseases.

Tables 7 demonstrates all causes of anemias in 668 patients including causes other than hemoglobinopathies. The most common etiology was hemoglobinopathies (34.28%) followed by iron deficiency (24.55%), and pregnancy (15.72%). The other causes of anemia were chronic diseases (13.47%), organ failure (4.49%), autoimmune diseases (3.29%), malignancy (2.1%), and unknown etiology (2.1%).

DISCUSSION

The hemoglobinopathies are generally divided into two types: the thalassemia (disorders of globin chain production) and the structural variants of hemoglobin such as hemoglobin S, E, C, etc. In addition, a combination of the aforementioned types can also be seen. Hemoglobinopathies are a wide group of inherited defects of globin chains. Mutations in globin genes result in quantitative defects of the globin chains resulting in an imbalanced synthesis of hemoglobin tetramers which end up in thalassemia. On the other hand, if globin gene mutations entail an abnormal globin protein, it produces variant hemoglobins.

A previous study performed on global epidemiology of hemoglobin disorders found that thalassemia and sickle cell disease are among the most common genetic diseases worldwide with an overall prevalence of more than one per cent in newborns with high mortality rates during early childhood [10]. The study also suggests that gene carrier risk of α or β-thalassemia, or hemoglobin S, C, D, or E is approximately 7% in pregnant females [10]. There might be a difference in the prevalence of hemoglobinopathies among different countries that can be partially attributed to non-availability of advanced diagnostic laboratory tests in resource-poor countries. Sickle cell anemia was found to be the most

Table 1. Anemia status based on hemoglobin levels.

Anemia status		Number of samples (n)	Percent (%)	Mean	Standard deviation
Hemoglobin (HGB)	severe	166	24.8	5.7	1.1
	moderate	305	45.7	8.5	0.9
	mild	197	29.5	11.0	0.5

Table 2. Distribution of MCV and MCH levels among the studied 668 subjects.

Parameters		MCH			Total number
		Low	Normal	High	
MCV	low	528	0	1	529
	normal	120	1	1	122
	high	6	7	4	17
Total		654	8	6	668

Table 3. Distribution of serum iron and ferritin levels among 528 subjects with microcytic hypochromic anemia.

Parameters		Ferritin			Total
		Low	Normal	High	
Serum Iron	low	137	63	32	232
	normal	60	129	70	259
	high	9	12	16	37
Total		206	204	118	528

Table 4. Descriptive statistics of sickle screen test among the studied subjects.

Sickle screen test	Number of samples (n)	Percent (%)
Negative	512	76.6
Positive	156	23.4
Total	668	100.0

prevalent hemoglobinopathy causing anemia in our study. We compared the results of this study with other studies conducted in Jeddah and on a national scale to know the frequencies of sickle cell trait and found it to be 5.7% [11], 4.2% [12], and 5.4% in Jeddah [3], 4.58% [13], 4.24% on a national scale, and 2.8% in the western region [14], much lower frequencies than in our study (9.73%). Sickle cell disease also showed a significant rise in prevalence compared to previous studies done on a national scale with frequencies of 0.26% [12], 0.24% [15], 1.4% [14], 0.38% [13], 0.27% and 0.08% in the western region [14] as compared to results of our study (9.43%).

Thalassemias are the most common congenital single-gene disorder worldwide and are most prevalent in malaria-endemic areas including the Middle East, the Mediterranean area, Southeast Asia, the Indian subcontinent, and Africa. A steady decline in the incidence of births of infants with β-thalassemia has been noticed in Mediterranean at-risk populations owing to extensive screening programs and prenatal diagnosis [15]. However, it is still a major clinical problem in other regions [16].

Table 5. Hematologic variables, biomarkers of iron status for normal Hb genotype (Hb AA) and the eight majors abnormal Hb variants for the studied subjects *.

Parameters		β-Thalassemia major	β-Thalassemia trait	Hb E/β	Hb S/β	Hb E	Hb SS	Hb AS	Hb H
Normal Values (mean ± SD)									
RBC (10^6/µL)		3.03 ± 0.86 *	4.8 ± 1.01 *	3.23 ± 0.71 *	3.72 ± 0.73	4.67 ± 1.33	2.77 ± 0.80 *	4.12 ± 1.03	4.6 ± 0.99
Male	4.8 ± 0.4								
Female	4.1 ± 0.5								
Hb (g/dL)		6.35 ± 1.31 *	8.99 ± 1.98	6.03 ± 1.45 *	7.87 ± 1.28	9.78 ± 2.15	7.62 ± 1.76 *	9.18 ± 2.21	7.79 ± 1.06
Male	15.2 ± 2.3								
Female	13.7 ± 1.8								
HCT (%)		19.92 ± 3.37 *	28.55 ± 5.73	19.23 ± 4.73 *	24.17 ± 3.48 *	30.78 ± 7.16	22.23 ± 4.83 *	28.18 ± 6.14	27.67 ± 4.11
Male	43 ± 3.0								
Female	39 ± 5.0								
MCV (fL)		67.58 ± 7.72 *	60.02 ± 6.95 *	59.43 ± 4.55	66.18 ± 9.16	66.8 ± 6.94	81.9 ± 10.55 *	69.63 ± 10.13	61.3 ± 7.13
Male	83.8 ± 5.6								
Female	84.1 ± 5.7								
MCH (pg)		21.38 ± 2.51	18.85 ± 2.52 *	18.65 ± 1.42	21.62 ± 3.85	21.85 ± 2.73	28.11 ± 4.39 *	25.37 ± 21.74	17.36 ± 2.72
Male	29.8 ± 2.0								
Female	28.9 ± 2.6								
RDW (%)		29.15 ± 4.06 *	18.55 ± 3.23	32.68 ± 1.93 *	22.35 ± 4.07 *	20.87 ± 7.44	21.09 ± 4.13 *	19.03 ± 4.81	27.2 ± 6.47 *
Male	13.2 ± 0.8								
Female	12.9 ± 0.7								
Retic (10^6/µL)		0.09 ± 0.11	0.07 ± 0.04	0.18 ± 0.08 *	0.2 ± 0.10 *	0.09 ± 0.07	0.3 ± 0.12 *	0.10 ± 0.10	0.19 ± 0.04 *
Male	1 ± 0.5								
Female	1 ± 0.5								

Table 5. Hematologic variables, biomarkers of iron status for normal Hb genotype (Hb AA) and the eight majors abnormal Hb variants for the studied subjects * (continued).

Hb AA		4 ± 1.00			8.64 ± 2.18			27.87 ± 6.19			72.24 ± 25.48			22.6 ± 12.15			18.49 ± 4.43			0.08 ± 0.08		
Parameters	Normal Values (mean ± SD)	RBC (10^6/µL)	4.8 ± 0.4	4.1 ± 0.5	Hb (g/dL)	15.2 ± 2.3	13.7 ± 1.8	HCT (%)	43 ± 3.0	39 ± 5.0	MCV (fL)	83.8 ± 5.6	84.1 ± 5.7	MCH (pg)	29.8 ± 2.0	28.9 ± 2.6	RDW (%)	13.2 ± 0.8	12.9 ± 0.7	Retic (10^6/µL)	1 ± 0.5	1 ± 0.5
			Male	Female		Male	Female		Male	Female		Male	Female		Male	Female		Male	Female		Male	Female

*** Values are arithmetic means ± SD. * Different from Hb AA, $p < 0.05$. RBC - red blood cell count, Hb - haemoglobin, HCT - haematocrit, MCV - mean cell volume, MCH - mean cell haemoglobin, RDW - red cell distribution width, Retic - reticulocyte.**

Table 6. Hemoglobin studies of 668 patients visited KAUH in Jeddah, Western of Saudi Arabia.

Hemoglobin Disorder	Number of samples (n)	Percent (%)
Normal hemoglobin (HbAA)	439	65.72
Sickle cell trait	65	9.73
Sickle cell anemia (HbSS)	63	9.43
β-Thalassemia trait	48	7.19
S/β-thalassemia	27	4.04
Hemoglobin H	7	1.05
β-Thalassemia major	6	0.90
Hemoglobin E	6	0.90
E/β-Thalassemia	4	0.60
Hemoglobin C	1	0.15
Hemoglobin D	1	0.15
Hemoglobin S/C	1	0.15
Total	668	100

Table 7. Prevalence of the underlying causes of anemia.

Underlying Cause	Number of samples (n)	Percent (%)
Hb disorders	229	34.28
Iron deficiency	164	24.55
Pregnancy	105	15.72
Chronic disease	90	13.47
Organ failure	30	4.49
Autoimmune	22	3.29
Tumor	14	2.10
NOS *	14	2.10
Total	668	100

*** NOS: not otherwise specified.**

Thalassemia is the most prevalent genetic disease globally while in Saudi Arabia, the most frequent hemoglobinopathy is Sickle cell and thalassemia [17] with a frequency as high as 45% in the eastern region counted for alpha thalassemia [14]. The symptomatic HbH disease subtype of α-thalassemia was included in our study with a prevalence of 1.0%, significantly higher than the previous study that was done in Jeddah with a prevalence of 0.15% [6]. In comparison to the previous studies to know the prevalence of beta-thalassemia trait showing frequencies of 3% [11], 3.22% [11], 4.69% in Jeddah [6], 1.29% [3], 1.85% on a national scale, and 1.0% in the western region [13], our study revealed a significant increase in its prevalence with 48/ 668 (7.2%) patients having the beta-thalassemia trait. Similarly, our results for the β-thalassemia major with the frequency of 0.9% also demonstrated a significant increase in comparison with previous studies done on a national scale with the prevalence of 0.07% [7,8], 0.05% as well as 0.02% in the western region [13] and 5.4% in Jeddah.

Our study also reported less prevalent types of hemoglobinopathies. Hemoglobin E was found in 6 (0.9%) patients while hemoglobin C and hemoglobin D had one patient each. There was not much difference noted in the previous study done in Jeddah, demonstrating a prevalence of 0.85% to hemoglobin E and 0.03% to hemoglobin C [6]. Four of our subjects had hemoglobin E combined with β-thalassemia (HbE/β) with a prevalence of 0.6%. Another subject of our study had sickle cell combined with hemoglobin C (HbSC) with a prevalence of 0.1%. A previous study indicated similar result with regard to HbSC, reporting 2 subjects with HbSC and they were both Saudis with West African ancestry [18]. The area with the highest prevalence of structural hemoglobin variants S and C is tropical Africa, but also very common in Saudi Arabia, Mediterranean countries, and Caribbean countries [10]. According to an estimate in 2010, the worldwide incidence of homozygous SS disease in newborns was over 300,000 and sickle cell trait incidence was above five million, with sub-Saharan Africa having the majority of these infants [19]. On the other hand, hemoglobin E is most prevalent in Southeast Asia with an estimated one million homozygotes for EE disease and 30 million carriers [19].

Multiple factors affect the incidence and prevalence of autosomal recessive disorders such as hemoglobinopathies and include population demographics and cultural characteristics. A significant level of genetic diversity in Arab countries stems from a large number of migrants from southeast Asia, Europe, and Africa [20] in the present-day genetic profile of the hemoglobinopathies. The higher prevalence of hemoglobinopathies in Saudi Arabia and other Arab countries can be attributed to customary consanguineous marriages especially first cousin unions constituting 25% of all marriages in many Arab countries [21,22]. Moreover, other factors contributing to the higher frequency could be a selective survival advantage against falciparum malaria, comparatively larger families with more than one affected child, and low resources of health services toward diagnosis and management of these disorders. Despite the high frequency of hemoglobinopathies, it has been challenging to render comprehensive healthcare services, presence of other pressing life-threatening disorders, the paucity of trained health professionals, and non-availability of data on the real magnitude, economic, and health burden of hemoglobinopathies. Furthermore, screening and care initiatives are deterred by the lack of awareness about risks of disease in addition to the cultural, religious, and legal limitations, such as fear of stigmatization within the community for families with genetic disorders and the religious and legal restrictions to abortions of affected fetuses.

The volume of available data for hemoglobinopathies in Arab countries is minimal. However, the published research articles in local or international journals provide a general insight into such provided facilities. The main service at the current time includes screening programs such as premarital screening for hemoglobinopathies. Moreover, Islamic teachings in medical genetics underscore the significance of education, counseling, and screening to deter the incidence of genetic disorders. The employment of genetics is not questionable with the exception of prenatal diagnosis and selective pregnancy termination [22-24]. Newborn screening for hemoglobinopathies enables early identification of the affected infants shortly after birth providing proper management and care prior to development of complications. For instance, early identification of sickle cell disease via neonatal screening aids to reduce the mortality rate by an early intervention of prophylactic pneumococcal vaccination and penicillin antibiotics. Similarly, the identification of carriers for sickle cell disease points out the possibility of having affected children so that future reproductive options and preventive measures can be undertaken. Newborn screening programs for sickle cell trait and disease are implemented in many Arab countries including Saudi Arabia that not only provide data on birth rates but also allows counselling of carrier parents along with prophylactic management of affected infants [25]. Moreover, preventive strategies are also involved in community-based care programs such as education and awareness campaigns and genetic counselling. Such measures have proven successful in different cultural settings leading to a decline in the number of newborns with these conditions [26].

A premarital screening program is among the most effective ways for prevention of certain acquired or inherited disorders. The main goal of this screening is to detect carriers for hemoglobinopathies so that they are informed about the prevention strategies and care before making informed decisions. The couple screening for hemoglobinopathies involves an initial measurement of mean corpuscular volume followed by hemoglobin electrophoresis. In Saudi Arabia, a national premarital screening for thalassemia and sickle cell anemia was started on a volunteer basis in 2004 in all provinces and in 2005 it became mandatory for all couples planning to

get married to be screened for hemoglobinopathies carrier status before the issuance of marriage certificates [27]. It was recommended to spread public awareness about genetic diseases even before individuals plan to get married through the curriculum in educational institutes and mass media messages. It is crucial to conduct PMS significantly in advance of the wedding plans in order to avoid the social embarrassment of wedding cancellation. So, the better option would be to introduce the screening during secondary school years [28].

Owing to the high prevalence of β-thalassemia and other hemoglobinopathies with an increased burden on health resources and society, it is mandatory to undertake a few effective measures to control its incidence. Prenatal diagnosis is considered to be an important option to prevent serious genetic disorders especially those at higher risk in the developed world. Screening with red cell indices for all pregnant women to detect more hemoglobinopathy carriers is recommended by the American College of Obstetricians and Gynecologists. If hemoglobinopathy is suspected on the basis of ethnicity (Middle Eastern, Mediterranean, African, West Indian descent, or Southeast Asian) or red cell indices showing a low MCV or MCH, the American College of Obstetricians and Gynecologists recommends a hemoglobin electrophoresis test in addition to a complete blood count. After the confirmation of fatal hemoglobinopathies, genetic counseling and selective termination would help curb the incidence of disease. However, there are several social, religious, and legal restrictions regarding the selective termination of pregnancy of an affected fetus in Saudi Arabia. A survey with a pre-structured questionnaire was conducted in Saudi Arabia in 2001 that involved families having children affected by a major hemoglobinopathy to evaluate their stance and acceptability of prenatal diagnosis and selective pregnancy termination. The study found that most of the recruited patients (81.3%) accepted prenatal diagnosis and the decision towards abortion was influenced by mainly by religious values [29]. On the other hand, pre-implantation of genetic diagnosis allows couples to avoid selective termination of an affected pregnancy [30].

CONCLUSION

Hemoglobinopathies are one of the major causes of anemia in Saudi Arabia presenting in significant numbers at the tertiary care hospital of Jeddah, Saudi Arabia. Our study demonstrated that more than one-third of anemic patients have these disorders with implications and burden on health authorities and services. High prevalence rates of hemoglobinopathies pose considerable challenges in Saudi Arabia requiring the institution of preventive national programs by establishing public health services not requiring sophisticated technical facilities. The strategies should take into consideration the unique prevalence data and cultural norms of each community, and the resources available. Community services that have demonstrated their effectiveness in reducing the prevalence rates of sickle cell disorders and β-thalassemia while simultaneously proposing appropriate management include NBS and PMS detecting haemoglobinopathies carriers coupled with genetic counselling. However, the matter of selective pregnancy termination of affected fetuses remains controversial. Some studies recommend hemoglobinopathy carrier screening of high school students, well ahead of marriage plans. Given the lack of knowledge and experience by several primary health care workers in implementing preventive and care interventions for hemoglobinopathies at the community level, educational programs and structured campaigns to educate and train health care providers ought to be mandated as a part of the national plan for the prevention of hemoglobinopathies. Diagnostic tests and management protocols for these disorders can be developed and updated with the knowledge of molecular defects and epidemiology of hemoglobinopathies in Saudi Arabia to curb the burden of genetic diseases.

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Declaration of Interest:
All authors declare that they have no conflict of interest associated with this publication.

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