Pattern of Hemoglobinopathies among Patients Attending PHC in Riyadh, Saudi Arabia

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Abstract

Background: Hemoglobinopathies pose a significant public health burden globally, with varying prevalence rates across different references. Understanding the pattern of hemoglobinopathies among patients attending primary healthcare centers (PHCs) is essential for guiding effective prevention and management strategies. Study Aim: To determine the pattern of hemoglobinopathies among patients attending PHCs in Riyadh, Saudi Arabia. Methodology: A cross-sectional study design was employed, involving 320 participants attending PSMMC PHCs in Riyadh. Demographic data and HGB types were collected, and Chi-square tests were conducted to test associations between demographic variables and HGB types. Results: The majority of participants were aged between 18 to 30 years (40.6%), with a nearly equal distribution of females (52.8%) and males (47.2%). All participants were of Saudi nationality. The most prevalent HGB genotype was S/S (63.1%), followed by A/S (36.6%). Significant associations were found between age groups and HGB types (χ^2 = 28.233, p < 0.001), with the S/S genotype more prevalent among younger participants. Gender did not show a significant association with HGB types. Conclusion: This study provides insights into the prevalence and patterns of hemoglobinopathies among patients attending PHCs in Riyadh, Saudi Arabia. The findings underscore the need for targeted screening and intervention programs tailored to specific age groups to effectively address hemoglobinopathies within the population.

Introduction

Background

Hemoglobinopathies represent a group of inherited blood disorders characterized by abnormalities in the structure or production of hemoglobin molecules [1]. These disorders are among the most prevalent genetic diseases worldwide, particularly in regions where consanguineous marriages are common. Hemoglobinopathies encompass a diverse range of conditions, including thalassemia and sickle cell disease (SCD), which pose significant health challenges due to their chronic nature and associated complications [1,2]. One of the major types of hemoglobinopathies is thalassemia, which results from mutations in the genes responsible for producing hemoglobin subunits. Beta-thalassemia, in particular, is characterized by reduced or absent synthesis of beta-globin chains, leading to an imbalance in the production of alpha and beta chains and subsequent hemolytic anemia [2,3]. Studies conducted in various regions have highlighted the
prevalence of beta-thalassemia traits among different populations. For instance, a study conducted in Bangalore, India, reported a prevalence rate of 8.6% among pregnant women attending antenatal care clinics [4].

Sickle cell disease (SCD) is another common hemoglobinopathy characterized by the presence of abnormal hemoglobin S (HbS) molecules. Individuals with SCD experience recurrent episodes of vaso-occlusive crisis due to the polymerization of HbS under hypoxic conditions, leading to painful episodes, organ damage, and increased susceptibility to infections [5,6]. The prevalence of SCD varies among different populations, with higher rates reported in regions with a high prevalence of malaria, such as sub-Saharan Africa. A study conducted in Lagos, Nigeria, reported a prevalence rate of 0.8% among infants screened for hemoglobinopathies [7].

The burden of hemoglobinopathies extends beyond the individual level to impact healthcare systems and economies, particularly in low- and middle-income countries where resources for comprehensive care and management are limited. Early detection and diagnosis of hemoglobinopathies are crucial for implementing preventive measures and providing appropriate interventions to reduce morbidity and mortality associated with these conditions [8-10]. Neonatal screening programs have been established in several countries to facilitate early identification of affected individuals and enable timely intervention.

In Saudi Arabia, hemoglobinopathies, particularly beta-thalassemia and sickle cell disease, are prevalent due to high rates of consanguineous marriages and the heterogeneous nature of the population [11]. Studies conducted in different regions of Saudi Arabia have reported varying prevalence rates of hemoglobinopathies. For example, a nationwide premarital screening program conducted between 2004 and 2007 reported a prevalence rate of 0.17% for beta-thalassemia and 0.25% for sickle cell trait among individuals undergoing screening [12].

**Study Aim**

This study aims to improve health care provided to the patients attending primary healthcare centers (PHCs) in Riyadh, Saudi Arabia.

**Study Objectives**

1. To determine the prevalence of different types of hemoglobinopathies, including sickle cell disease, thalassemia, and other variants, among patients attending PHCs in Riyadh.
2. To describe the demographic characteristics (age, gender, nationality) of patients diagnosed with hemoglobinopathies in the study population.
3. To test any associations between demographic variables (age, gender) and specific types of hemoglobinopathies among patients attending PHCs.

**Methodology**

**Study Design**

A cross-sectional study design was employed to investigate the prevalence and pattern of hemoglobinopathies among patients attending primary healthcare centers (PHCs) in Riyadh, Saudi Arabia.

**Study Setting**

The study was conducted at Prince Sultan Military Medical City (PSMMC) primary healthcare centers in Riyadh, Saudi Arabia. These centers serve as the primary point of contact for healthcare services for eligible patients in Riyadh and represent a diverse population base.

**Participants**

A total of 320 participants attending PHCs in Riyadh were included in the study. Participants were selected through convenience sampling based on their willingness to participate and availability during the study period. Inclusion criteria comprised individuals of all age groups who were tested for hemoglobin electrophoresis during the study period.

**Data Collection**

Data collection was conducted over a specified period, during which demographic information and hemoglobin (HGB) types were recorded for each participant. Information regarding age, gender, and nationality was collected as part of the demographic profile. Hemoglobin types, including A/F, A/S, and S/S, were determined through laboratory analysis of hemoglobin electrophoresis.

**Statistical Analysis**

Data analysis was performed using appropriate statistical methods to examine the prevalence and distribution of hemoglobinopathies among the study population. Descriptive statistics, such as frequencies and percentages, were calculated for demographic variables and hemoglobin types. Chi-square tests were employed to assess associations between demographic variables and hemoglobin types, with significance set at p < 0.05. Statistical analysis was conducted using SPSS version 26 software.

**Ethical Considerations**

Ethical approval for the study was obtained from the Institutional Review Board (IRB) of Prince Sultan Military Medical City. Ensuring confidentiality of patient data. The study adhered to ethical principles outlined in the Declaration of Helsinki and other relevant guidelines.

**Results**

Table 1 provides a comprehensive overview of the demographic profile and the prevalence of different HGB types among the participants. The majority of participants fell within the age range of 18 to 30 years (130 participants, 40.6%), followed by those less than
18 years old (89 participants, 27.8%). Gender distribution indicated a near-equivalent representation, with 169 females (52.8%) and 151 males (47.2%). Notably, all participants belonged to the Saudi nationality. Regarding HGB types, the S/S genotype was the most prevalent (202 participants, 63.1%), followed by A/S (117 participants, 36.6%), with only one participant exhibiting the A/F genotype (0.3%).

Table 2 further examines the relationship between demographic variables and HGB types through Chi-square tests. Age groups exhibited statistically significant associations with HGB types ($\chi^2 = 28.233$, $p < 0.001$). Participants aged less than 18 years predominantly displayed the S/S genotype (62 participants, 69.7%), while those aged 18 to 30 years showed a balanced distribution between A/S and S/S genotypes. Similarly, gender did not significantly associate with HGB types ($\chi^2 = 1.343$, $p = 0.511$), with both females and males demonstrating comparable distributions across HGB genotypes.

![Figure 1: Boxplots Representing Age Distribution Across Different Types of HGB (n=320)](image)

### Table 2: Characters of the Included Sample in Association with Types of HGB (n=320)

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Type of HGB</th>
<th>$\chi^2$</th>
<th>P-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age, y</td>
<td>A/F</td>
<td>A/S</td>
<td>S/S</td>
</tr>
<tr>
<td>Less than 18</td>
<td>0 (0%)</td>
<td>27 (30.3%)</td>
<td>62 (69.7%)</td>
</tr>
<tr>
<td>18 to 30</td>
<td>0 (0%)</td>
<td>38 (29.2%)</td>
<td>92 (70.8%)</td>
</tr>
<tr>
<td>31 to 40</td>
<td>0 (0%)</td>
<td>31 (44.9%)</td>
<td>38 (55.1%)</td>
</tr>
<tr>
<td>41 or more</td>
<td>1 (3.1%)</td>
<td>21 (65.6%)</td>
<td>10 (31.3%)</td>
</tr>
<tr>
<td>Gender</td>
<td>Female</td>
<td>0 (0%)</td>
<td>64 (37.9%)</td>
</tr>
<tr>
<td>Male</td>
<td>1 (0.7%)</td>
<td>53 (35.1%)</td>
<td>97 (64.2%)</td>
</tr>
<tr>
<td>Nationality</td>
<td>Saudi</td>
<td>1 (0.3%)</td>
<td>117 (36.6%)</td>
</tr>
</tbody>
</table>

**Discussion**

Hemoglobinopathies represent a significant public health concern globally, particularly in regions where the prevalence is high. These genetic disorders, including sickle cell disease and various forms of thalassemia, can lead to severe health complications and pose considerable burdens on healthcare systems [1-3]. In Saudi Arabia, as in many other countries, hemoglobinopathies are a major health challenge, necessitating a thorough understanding of their prevalence and distribution among the population.
prevalence and patterns to inform effective prevention and management strategies [11,12]. This study aimed to investigate the pattern of hemoglobinopathies among patients attending primary healthcare centers (PHCs) in Riyadh, Saudi Arabia. The findings of this study revealed several important insights into the demographic characteristics and distribution of hemoglobin (HGB) types among the study participants. The majority of participants were between the ages of 18 to 30 years, with a near-equivalent representation of females and males. Notably, all participants were of Saudi nationality. Regarding HGB types, the S/S genotype was the most prevalent, followed by A/S, with minimal occurrences of the A/F genotype. Further analysis revealed significant associations between age groups and HGB types, while gender did not show significant associations.

The prevalence and distribution of hemoglobinopathies observed in this study align with findings from previous research. For instance, studies conducted in different regions of Saudi Arabia have reported the predominance of the S/S genotype observed in our study is consistent with the high prevalence of sickle cell disease reported in Saudi Arabia and other countries with a high prevalence of malaria [18,19]. The significant association between age groups and HGB types underscores the importance of considering age-related factors in the assessment and management of hemoglobinopathies. Similar associations have been documented in previous studies, suggesting that the distribution of HGB types may vary across different age cohorts [20,21]. This finding emphasizes the need for targeted screening and intervention programs tailored to specific age groups to effectively address hemoglobinopathies within the population. Contrary to some expectations gender and nationality did not exhibit significant associations with HGB types in our study. While previous research has suggested potential gender and ethnic disparities in the prevalence of hemoglobinopathies, the lack of significant associations in our study may reflect the homogeneous nature of the sample population, composed entirely of Saudi nationals [22,23]. However, it is essential to recognize that genetic and sociodemographic factors may still influence the prevalence and distribution of hemoglobinopathies, warranting further investigation in diverse populations. Comparing our findings with those of previous studies highlights both similarities and differences in the prevalence and patterns of hemoglobinopathies across different populations. The predominance of the S/S genotype in our study mirrors findings from other hemoglobinopathy-endemic regions, emphasizing the global burden of sickle cell disease and the need for targeted preventive measures [24,25]. Similarly, the association between age groups and HGB types observed in our study corroborates evidence from existing literature, suggesting age-related variations in the prevalence of hemoglobinopathies [26,27]. However, discrepancies in the association between gender, nationality, and HGB types underscore the complex interplay of genetic, environmental, and sociodemographic factors influencing the distribution of hemoglobinopathies. While some studies have reported gender and ethnic disparities in the prevalence of specific HGB types, our findings suggest a more nuanced understanding of these relationships, highlighting the importance of population-specific considerations in hemoglobinopathy research [28,29].

Conclusion

In conclusion, this study provides insights into the prevalence and patterns of hemoglobinopathies among patients attending PHCs in Riyadh, Saudi Arabia. The findings underscore the need for targeted screening and intervention programs tailored to specific age groups and populations to effectively address the burden of hemoglobinopathies within the region. By comparing our findings with existing literature, we contribute to a deeper understanding of the global epidemiology of hemoglobinopathies and inform evidence-based strategies for prevention, management, and healthcare delivery in hemoglobinopathy-endemic regions.

References


