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Research Article

Clinicohematological Characteristics, Types and Frequency of Hemoglobinopathies; A Nonhospital Based Laboratory Experienced from Pakistan

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Abstract

Introduction: Thalassemia is a genetic disease which has autosomal recessive pattern of inheritance with carrier rate of thalassemia is 5-7% in Pakistan. The study was planned to observe the types, frequency and detailed clinicohematological of hemoglobinopathies in the Karachi, the largest cosmopolitan city of the country.

Materials and Methods: An observational and cross-sectional study conducted during the period of September 2020 to February 2022 at Dr. Essa Laboratory and Diagnostic center. A total of 1365 participants were included. CBC was performed using XP-100 and Hb Electrophoresis was done using Bio-Rad D10 analyzer. All parameters were calculated in mean, standard deviation and percentages using SPSS version 23.

Results: Among 1365 participants, the Iron deficiency anemia (IDA) was more common in patients 971(71%) followed by thalassemia minor 276 (20%), thalassemia major 90 (7%), HbD disease 20 (1.46%) and HbE disease 08 (0.5%). The most common presenting complain was weakness observed in 933 (68.3%) patients followed by vertigo in 230 (16.8%) and body pain in 102 (7.47%) patients. A significant low level of Hb 5.9 ± 2.95 and MCV 17.3 ± 4.9 were observed in Thalassemia major patients with p = < 0.001.

Conclusion: Although thalassemia is a genetic disease, but higher frequency was found in our population with IDA and thalassemia minor. Screening before marriages and adequate measures especially prenatal diagnosis should be performed to reduce the possibility of Hemoglobinopathies. However, this study has been done on small number of sample size in one large cosmopolitan city of Karachi and requires evaluating further with large number of sample size within different areas of Pakistan.

Keywords: Clinicohematological; Frequency; Hemoglobinopathies; Pakistan

Abbreviations

Hb: Hemoglobin; MCV: Mean Cell Volume; MCH: Mean Cell Hemoglobin; MCHC: Mean Cell Hemoglobin Concentration; TLC: Total Leucocytes Counts

Introduction

Thalassemia is a genetic disease which has autosomal recessive pattern of inheritance [1]. In all over the world approximately 6000 thalassemia neonates are born with the 3% frequency of beta thalassemia carrier [2]. The distribution of specific disorders

varies geographically and by community. The estimated carrier rate of thalassemia is 5- 7% in Pakistan as the disease is result of consanguineous marriages and huge number of such marriages persist in third world country like Pakistan due to socio-cultural issues [3,4]. The clinical presentation includes weakness, pallor and failure to thrive in early phase of disease, required frequent blood transfusions resulting in iron overload many metabolic complications [5]. In third world country like Pakistan, the screening and diagnosis of thalassemia is limited due to lack of knowledge, sociocultural issue, and financial resources. For the diagnosis the CBC is considered as the first and the most important laboratory test for the thalassemia carriers and diseased patients [6]. For the carriers of both α- or β-thalassemia carriers, patient are present with microcytic hypochromic parameters with or without anemia requiring for the exclusion of IDA [7]. The diagnosis includes the red cell indices with morphology with the measurement of Hb variants by HPLC. The determination of HbA2 is the most accurate detection carriers with simple method by the cation exchange HPLC and capillary electrophoresis. The highest levels of fetal hemoglobin with the absence of HbA represents classical picture of B thalassemia major [8]. The study was planned to observe the types, frequency and detailed clinico hematological characteristics in frequency distribution of hemoglobinopathies in the Karachi, located in the southern part of Pakistan, the largest cosmopolitan city of the country.

Materials and Methods

An observational and cross-sectional study conducted during the period of September 2020 to February 2022 at Dr. Essa Laboratory and Diagnostic center. A total of 1365 participants were included. Clinical details were recorded through Hb electrophoresis history form. Inclusion of all participants was voluntarily. The 3cc venous blood samples were taken to the K2-EDTA tubes. CBC was performed using XP-100 (Sysmex Corporation, Kobe, Japan) and all red cell, white cells and platelets parameters were recorded. The Hb Electrophoresis was done using Bio-Rad D10 analyzer (Bio-Rad Laboratories, Inc., Hercules, CA).

Statistical analysis

The parameters were calculated as in mean, median, standard deviation and percentages and chi square was used to find difference between the studied groups by using SPSS version 23 with significant difference of <0.001.

Results

A total of 1365 participants were enrolled in the study. The Iron deficiency anemia (IDA) was more common in patients 971(71%) followed by thalassemia minor 276(20%), thalassemia major 90(7%), HbD disease 20(1.46%) and HbE disease 08 (0.5%). (Figure 1) The most common presenting complaint was weakness observed in 933(68.3%) patients followed by vertigo in 230(16.8%) and body pain in 102 (7.47%) patients. (Figure 2) The mean age of patients was 22.41 ± 9.89. Females were higher in number as 312 (%) and males were 150 (%). The hematological characteristics revealed significant low levels of Hb5.9 ± 2.95g/dl in BTM patients as compared with other groups. The red cells indices including MCV and MCH were significantly lower in all studied groups. The platelet levels were significantly raised in IDA patients 433 ± 48 in IDA group and the lowest counts were observed in BMT 130 ± 68*10^3/µL predicting hemolysis in BTM patients (Table 1).

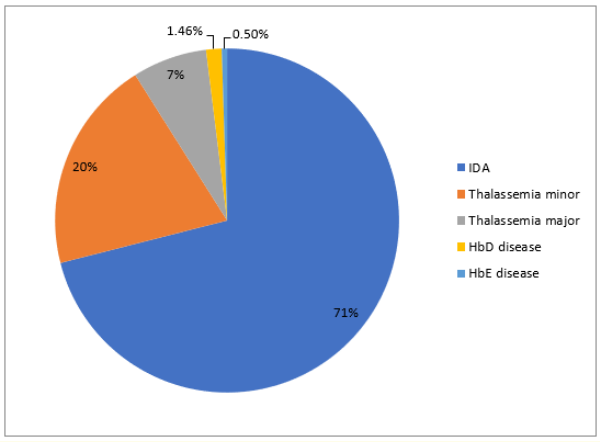

Figure 1: Frequency of IDA and other Hemoglobinopathies.

Discussion

Thalassemia is an autosomal recessive disease. The main purpose of this study was to evaluate the frequency of β-thalassemia and other hemoglobinopathies in the largest non-hospital-based laboratory with various ethnic origins in the cosmopolitan city of Karachi, Pakistan. Among the study participants, a total of 815 (59.7%) females and males were 550 (40.2%) were included which are dissimilar with the study of Ansari., *et al.* as highest frequency of male patients (53%) were included [4]. The means age was.

22.41 ± 9.89 which is less than the study of Mondal as mean age was 25.8 years [9]. The Anemia was common in all patients. The most common entity of anemia was IDA (71%) and 29% had presented with Hb disorder which is more than the previous studies [9,10]. Among the Hb abnormalities, the detection of β thalassemia trait was the most common with the frequency of 20% as observed in other studies [11-14]. The frequency of B thalassemia major was 7% which is more than the studied previously [14]. The frequency of other variants including HbD (1.46%) and HbE (0.50%) was less as compared with previous studies [15,16]. (Figure 2) The current findings are suggestive of multiple Hemoglobinopathies due to lack of awareness for the inheritance of thalassemia. The most common presenting complaint was weakness (68.3%) followed by vertigo (16.8%) and body pain. (7.4%) (Figure 2) Among the red cell indices, significant low levels of Hb 5.9 ± 2.95 and MCV 17.3 ± 4.9 were observed in Thalassemia major patients with p = < 0.001 as compared with the study of Shaista., *et al*. [17]. (Table 1).

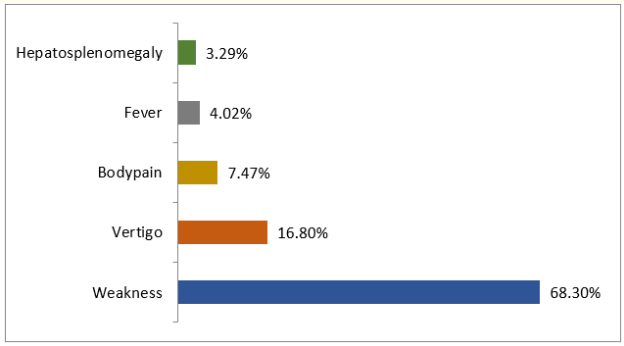

Figure 2: Frequency of symptoms observed.

Parameters	IDA	Thal Minor	Thal Major	HbD Disease	HbE Disease	P-value
Hb(g/dl)	7.3 ± 4.9	6.6 ± 3.24	5.9 ± 2.95	9.94 ± 4.94	8.8 ± 4.12	<0.001
MCV (fl)	68 ± 4.07	71 ± 6.2	65 ± 5.1	76.6 ± 7.07	74 ± 7.2	<0.001
MCH (pg)	19. ± 5.65	16.1 ± 6.7	17.3 ± 4.9	20.1 ± 4.3	22.1 ± 5.6	<0.001
MCHC(g/dl)	26 ± 4.94	27 ± 5.1	25 ± 4.0	30.1 ± 4.9	28 ± 3.8	0.63
TLC (*10^3/µL)	9.98 ± 2.1	7.6 ± 1.1	4.2 ± 2.2	10 ± 1.0	8.8 ± 2.0	<0.001
Platelets (*10^3/µL)	433 ± 48	320 ± 51	130 ± 68	350 ± 36.6	260 ± 55.8	<0.001

Table 1: Hematological characteristics of studied population.

For the prevention of thalassemia, genetic counseling is valuable. In the present study, all patients with thalassemia trait were counseled for the disease mode of inheritance ethically with and privacy of every participant was maintained. In Pakistan, many thalassemia prevention centers are available now for the diagnostic facilities, treatment, and genetic counseling. On a large scale, awareness session had been conducted as prenatal diagnosis is major step for the prevention. In western countries and Mediterranean region, a successful decline in thalassemia trend has been observed due to the implementation of many screening programs [18]. Several screening programs have been started in Pakistan which includes pre marriage screening, extended family. screening of index child and mass screening. A huge number of public programs are needed along with screening programs for the achievement of mass education through print and electronic media, seminars, symposiums for medical and non-medical professional including people of every age groups which would impact in reducing the frequency of thalassemia carriers and disease patients as the expenditures for the bone marrow treatment of thalassemia major is very much costly which is not affordable to the most of the people living in Pakistan.

Conclusion

The early detection of hemoglobinopathies and variants can be done by simple, less time consuming and accurate method.

of HPLC. The high frequency of thalassemia trait is still a burden to the population. Thus, premarital screening can prevent Hemoglobinopathies. In countries like Pakistan, mass education and family studies would be helpful in the evaluation of thalassemic carriers. Moreover, a large-scale studies are needed to formulate preventive and appropriate treatment strategies regionally.

Declaration of Conflicting Interests

The author(s) declare no potential conflicts of interests with respect to the authorship and/or publication of this article.

Acknowledgment

A.A collected the samples and data, literature review, statistical analysis, contributed to paper writing and finalized the manuscript. NFE and FEA supervised the project and critically reviewed the manuscript. All authors reviewed the manuscript for important intellectual content and approved the final manuscript.

Contributions

We are very thankful to all study participants who participated voluntarily. The staff of hematology department for performing CBC of all samples. Senior most technologists Yamin Baig for performing HPLC of all samples.

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