HAEMOGLOBANOPATHIES IN SOUTHERN AREAS OF KHYBER PAKHTUNKHWA

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ABSTRACT

Objectives: To determine the frequencies of various types of haemoglobanopathies in southern areas of Khyber Pakhtunkhwa.

Material and Methods: This cross sectional study was conducted in Department of Pathology, Gomal Medical College from January 2014 to October 2014. Relevant information's notede on a questionnaire prepared in accordance with the aims and objectives of the study.

Results: A total of 62 patients underwent Hb electrophoresis in Department of Pathology Gomal Medical Collge, Dera Ismail Khan. 27(43.5%) were females and 35 males (56.5%). Majority of the patients 37(59.7%) were in the age range 3 month to 5 years. We received patients from age 3 month to 60 years age for knowing types of haemoglobanopathies. The distribution of patients was 49(79%) patients DI Khan, Tank 10(16%), Lakki Marwat 2(3.2%) and South Wazirstan 1(1.6%). The Haemoglobin of all patient as recorded from the peripheral smear. It was observed that 43(55%) patients had Hb < 8g/dl and were candidates for blood transfusion. Of the above group 17 had Hb ranging from 2.2 to 5g/dl (severely anemic). The Distribution of various haemoglobanopathies were; Beta thallassemia major 16(25.8%), Beta Thalassemia trait 7(11.3%), Sickle cell disease 6(9.7%), Sickle cell trait 2(3.2%), Sickle cell/Beta thalassemia (double heterozygosity) 2(3.2%) and HbE/ Beta Thalassemia (Double heterozygous 1(1.6%). 28(45.2%) had normal Hb Electrophoresis reports.

Conclusion: Haemoglobanopathies are common in southern areas of Khyber Pakhtunkhwa. Beta thalassemia major is a major haemoglobanopathy. Sickle cell diseases are also prevalent in this region especially in the Sherani, Baytani and Ustrana tribes of Dera Ismail Khan.

Key Words: Haemoglobanopathies, Thallasemia, sickle cell disease.

INTRODUCTION

Hemoglobinopathies are hereditary hematological disorders that results from structural abnormalities in one of the globin chains of the haemoglobin molecule[1]. Hemoglobinopathies are inherited in families as autosomal co-dominant traits. Sickle-cell disease and thallassemia are common hemoglobinopathies. It is reported that 7% of world's population (nearly 420 million) are sickle cell carriers, of those 60% carriers and 70% pathological cases reported from Africa.[2] Thalassemias usually results from underproduction of normal globin proteins, that is through mutations in regulatory genes for the specific protein molecule. The two conditions thalassemia nad sicle cell disease are important hemoglobinopathies. Beta-thalassaemia major is a common health problem in Pakistan. It is the very prevalent genetically transmitted hematological disorder with a carrier rate of 5-8%; about 5000 children are diagnosed every year in Pakistan[4]. In the heterozygous t type the beta thalassemia trait (ie, thalassemia minor) presents with mild to moderate hypochromic microcytic anemia. In developing countries the haemoglobinopathies like (thalassaemias and sickle-cell disorder) are common and they deal it by developing convenient model for prevention and genetic approach to control these diseases. It is reported that about 250 million people (4.5% of the total world population) carry a potentially pathological gene for causing haemoglobinopathies.[5] Sickle-cell disease (SCD) results when a person inherits two abnormal genes, one from each parents. Several subtypes of sickle cell hemoglobin exist, which depends on the exact mutation in haemoglobin gene. A person with a single abnormal gene does not present with symptoms of the disease and is called sickle-cell trait.[6] Hemoglobin (Hb) E, is a common Hemoglobin variant. It is found in Southeast Asia and is associated with beta thalassemia phenotype double heterozygosity (Hb E-Beta thallasemia double heterozygous), and this is included in the beta thalassemia type of diseases.[7]

Present study was therefore designed as to determine the frequencies of various types of haemoglobanopathies in southern areas of Khyber Pakhtunkhwa.

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MATERIAL AND METHODS

This cross-sectional study was conducted in the

Department of Pathology, Gomal Medical College, D.I. Khan, Pakistan from January 2014 to October 2014. A sample of 62 patients was collected by non probability convenience sampling. The study was carried out on the patients who were referred for the Hb electrophoresis from various health care facilities of the southern areas of Khyber Pakhtunkhwa. All patients referred for investigation for the cause of anemia or the purpose of screening was included irrespective of age and sex. Patients having blood transfusion in the last four weeks were excluded.

Detailed history regarding age, sex, race, family history, caste, blood transfusion history was recorded. Clinical examination was done for splenomegaly in all patients. About 2 to 3 ml of venous blood was taken from all patients in EDTA tube. Automated Hematology Analyzer wth specification (Nihon Kohden, Tokyo) readings were taken to determine cell counts and red cell indices like (RBC count, HB%, Hematocrit, Mean Cell Volume, Mean Corpuscular Hemoglobin, and Mean Corpuscular Hemoglobin Concentration). Peripheral blood films were also prepared for red blood cells and white bold cell morphology and any other related findings like hypochromasia etc.

Hemolysate prepared by adding distilled water and carbon tetrachloride to the sample of the blood. Hb electrophoresis was performed on Wealtech, machine, USA. Current flow was adjusted as per instruction of the manual of device. Cellulose acetate strip was used for application of hemolysate and different bands of hemoglobin were noted for reporting. All the results were noted and reported after comparing with the normal control on the Cellulose acetate strip.

As we know that HbD band appears due to HbD disease as well as sickle cell disease, so to differentiate in between the above two disorders sickling test was performed. Gender and age were demographic variables in present study while type of hemoglobinopathies and district wise catagorization were research variables. These all were analyzed as in frequency (%). Data entered in MS-Excel programme 2010 and analyzed in pi tables and then percentages were calculated using formulas on the pi-tables.

RESULTS

A total of 62 patients were selected in the present study conducted in the department of Pathology Gomal Medical collge Dera Ismail Khan. 27(43.5%) were females and 35 males (56.5%) Table 1. Majority of the patients 37(59.7%) were in the age range 3 month to 5 years. We received patients from age 3 month to 60 years age for knowing types of haemoglobanopathies. mainly patients were in childhood age and were referred by the respective physiscian for knowing the cause of anemia or haemoglobinopathy (Table 2).

This study included patients from the southern areas of the province of Khyber Pakhtunkhwa. The dis-

Table 1: Gender distribution of patients. (n=62)

Gender	No. of patients & percentage
Female	27(43.5%)
Male	35(56.5%)

Table 2: Age wise categorization of patients (n=62)

Age range	No. of patients & percentage
3m-5 year	37(59.7%)
5-10 years	3(4.8%)
11-15	6(9.7%)
16-20	6(9.7%)
21-25	4(6.5%)
26-30	4(6.5%)
40-45	1(1.6%)
55-60	1(1.6%)

Table 3: Area wise distribution of the patients (n=62)

Address	No. of patients & percentage
D.I. Khan	49(79.0%)
Lakki Marwat	2(3.2%)
South Waziristan Agency	1(1.6%)
Tank	10(16.1%)

Table 4: Haemoglobin of patients. (n=62)

Hb% Range	No. of patients & percentage
2.2-5.2g/dl	14(22.6%)
5.2-8.2 g/dl	20(32.3%)
8.2-11.2 g/dl	17(27.4%)
11.2-14.2 g/dl	9(14.5%)
14.2-17.2 g/dl	2(3.2%)

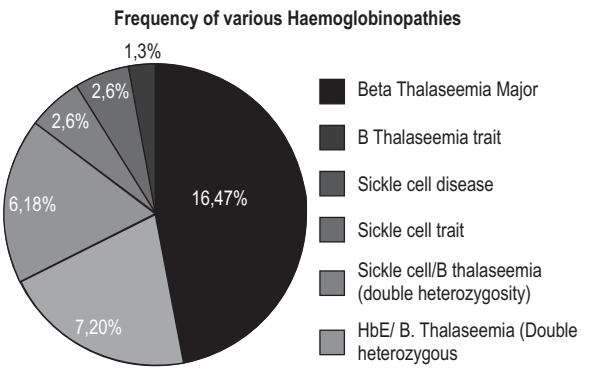

Figure 1: Frequency of various Haemoglobinopathies. (n=34)

tribution of patients was 49(79%) patients DI Khan, Tank 10(16%), Lakki Marwat 2(3.2%) and South Wazirstan 1(1.6%) Table 3.

As all these patients were anemic and referred for knowing the type of haemoglobinopathy. Many elder patients were also received for the purpose of knowing the carrier state or vice versa. The Haemoglobin of all patients as recorded from the peripheral smear. It was observed that 43(55%) patients had Hb<8g/dl and were candidates for blood transfusion. Of the above group 17 had Hb ranging from 2.2 to 5g/dl (severely anemic) Table 4.

Hb electrophoresis is done for knowing the type of Hb disorders. We observed that the distribution of various haemoglobanopathies were; Beta thallassemia major 16(25.8%), Beta Thalassemia trait 7(11.3%), Sickle cell disease 6(9.7%), Sickle cell trait 2(3.2%), Sickle cell/Beta thalassemia (double heterozygosity) 2(3.2%) and HbE/ Beta Thalassemia (Double heterozygous 1(1.6%). 28(45.2%) had normal reports (Figure 1).

DISCUSSION

Beta thalassemia major is a public health issue in our country. As all hemoglobinopathies are autosomal recessive disorders but due to our culture and taboos and giving preference to male children, male patients are referred more than females to the tertiary care hospitals. In present study 27(43.5%) patients were females and 35 males (56.5%) male to females ratio of 1.3:1. In another study conducted reported from abroad covering 667 patients 52.3% males and 47.7% female, also reported male predominance in their findings[8]. The reason of preponderance of males over females is the same as given in our study i.e more health care taken for males over females.

Sickle cell disease (SCD) remains a relatively common type of haemoglobinopathies in southern parts of Pakistan. It was found from the present study that it is more common in the southern areas of the province. In D.I. Khan district still it is more common in the tribes of the Baytani, Sherani and Ustrana (Storiani). One common justification for the prevalence of SCD in these tribes is the prevalence of malaria in these tribes. Sickle type of haemoglobin is more resistant to malarial parasite. Researchers have reported that sickle cell hemoglobin confers a survival advantage against malarial parasite[9]. Another study from Pakistan reported that Balochistan has the highest number of (SCD) patients 140. They concluded that sickle cell disorders are prevalent in southern belt of the country to a significant extent, starting from the tribes of southern part of the Khyber Pakhtunkhwa joining the areas of Balochistan[9]. We observed SCD in 10% while sickle cell trait in 3% of the sampling[10].

In present study the distribution of various haemoglobanopathies were; Beta thallassemia major 16(25.8%), Beta Thalassemia trait 7(11.3%), Sickle cell disease 6(9.7%), Sickle cell trait 2(3.2%), Sickle cell/Beta thalassemia (double heterozygosity) 2(3.2%) and HbE/ Beta Thalassemia (Double heterozygousity) 1(1.6%). 28(45.2%) had normal reports. In another study from Pakistan they reported that trait was detected in all ethnic groups with an overall prevalence rate of 5.4% (27/500) in their sampling. Pakhtuns had significantly (P <0.02) higher trait prevalence rate (7.96%) than those of Punjabis (3.26%)[11]. Another study from Punjab showed 61% of the subjects studies had haemoglobinopathies. Frequency of beta-Thalassemia trait was on the top followed by beta-Thalassemia major, HbE trait, HbD Punjab and Hb intermedia[12]. Still another study reported that thalassaemia trait (25.6%) was on the top followed by thalassaemia major (1.4%) and HbS or HbD (1.4%)[13].

In present study Sickle cell/Beta thalassemia (double heterozygosity) was noted in 2(3.2%) patients. The occurrence of HbS- β thalassemia due to inheritance of both the defective genes. The clinical presentation of HbS- β thalassemia are extremely changing, ranging from a asymptomatic type to a severe disease similar to homozygous type of sickle cell disease.[14]

We observed HbE/ Beta Thalassemia (Double heterozygousity) 1(1.6%). HbE itself presents with mild clinical picture, in even heterozygous or homozygous states,. But its association with other haemoglobinopathies, such as HbE/beta Thalassaemia double heterozygousity gives rise to a severe disease that is transfusion dependent thalassaemia syndrome[15]. Double heterozygosity thalassemia syndromes may present with severe clinical disease.

CONCLUSION

Haemoglobanopathies are common in southern areas of Khyber Pakhtunkhwa. Beta thalassemia major is a major haemoglobanopathy. But sickle cell diseases are also prevalent in this region.

Every child with sever anemia and families of patients with haemoglobanopthies must undergo screening to prevent chances of haemoglobanopathies. If these measures are not taken in near future, haemoglobinopathies will become endemic in this part of the country.

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AUTHOR'S CONTRIBUTION

Following authors have made substantial contributions to the manuscript as under:

Hussain J: Collection of data.

Khan H: Data analysis.

Ali SA: Article formulation.

Jan MA: Reviewing and editing.

Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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