Research Article

Spectrum of Thalassemia and Hemoglobinopathies in Pakistan: A Single Center Study

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Abstract

Background: Haematologists around the world have been striving hard to identify the spectrum of bleeding diathesis. The analysis of different bleeding disorders help in better management and outcomes of these patients. The rationale of the study was better cure and management of these diagnosed cases.

Objective: The study aimed to identify the prevalence and classify haemoglobinopathies and thalassemia in a major tertiary care center of capital province Punjab.

Method: The study was based on a cross sectional survey technique. The study was conducted in a retro-spective domain. We selected a sample of around 12,000 patients for our study with purposive non-probability sampling. The sample size comprised of 12000 patients presenting with anaemia on a complete blood count. Out of 12000 samples collected, 357 samples were rejected because of clotted/haemolized samples, rest 11643 samples were included in the study. The duration selected for this study started from 1st January 2019 to 1st January 2020 (1 year study period). The venue for collection of blood samples was the institute of pathology, Chughtai lab, Lahore.

Result: A total of 11643 samples received for Hb electrophoresis. Out of these 11643 cases 9145 samples turned out to be normal. 2498(21.4%) cases were abnormal results on electrophoresis. The percentages of all the hemoglobinopathies were calculated. Out of 2498 cases 61.2% had beta thalassemia trait, 26.5% of the population sample had beta thalassemia of major type, however the HbD trait was identified to have a value of 5.2%, HbD homozygous 0.3%, sickle trait .8%, sickle cell disease 0.7%, HbE trait 0.6%, delta/beta thalassemia 0.6%, sickle/beta thalassemia 2.3%,HbE /beta thalassemia 0.4%, HbH disease 0.2%, and other rare cases constituted 0.3%. these rare cases included one case of homozygous HbE, one case of HbC/ beta thalassemia, 3 cases in bart zone, two cases in J zone and one single diagnosis of sickle thalassemia was detected.

Conclusion: The conclusion of the study was very staggering as the trait of thalassemia (beta type) was toping the differential diagnosis. The second in differential was beta thalassemia of major type. Haemoglobinopathies other than above included the trait of HbD , sickle beta thalassemia, the trait of sickle and sickle cell disease. Heterozygotes like delta/beta, HbE/ beta thalassemia were less common.

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Keywords | Thalassemia, Hemoglobinopathies, Bleeding

Introduction

The term used for abnormal production of haemoglobin is called Hemoglobinopathies. Most of these blood disorders are inherited genetically. The estimated percentage of this blood disorder in the population of world varies around 5% approximately. The prevalence of haemoglobin production disorder in infant population is around 12,000 births per annum. So, an overall carrier rate of haemoglobinopathies is around 42 million people per year.[1]

The emerging trend of migration in human population and a relative tendency of high cognate marriages in under deve-

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https://doi.org/10.21649/jspark.v3i1.376

loped countries have both attributed to an ever-increasing trend of haemoglobinopathies.1 Many blood disorders are transferred as inheritance. Thalassemia is a blood disorder that is transferred from parents to their younger children. In Thalassemia the blood is deficient of a protein called haemoglobin. The patho-physiology of thalassemia involves abnormal synthesis of the polypeptide globin chain.[2] The two variants of thalassemia are (alpha) chain abnormality and (beta) chain abnormality thalassemia.[2]

The two most important haemoglobinopathies are thalassemia and sickle cell disease. The commonest trait of thalassemia in Pakistan is (beta) chain abnormality thalassemia. The average rate of this blood disorder is 5-8%. The per annum rate of diagnosis for thalassemia (beta) type in newborns is 5000 effected children per year.[4] The worldwide statistical analysis for haemoglobinopathies show a percentage of 4.5% of the total population of the world. Which counters to 250 million people carry a mutated gene causing haemoglobinopathies.[5]

The second most important inherited trait of blood disorder is a sickle cell disease. The sickle cell disease is caused by inheritance of two mutated gene, both from the child's mother and father. If the child carries only one mutated gene, it is called SICKLE CELL TRAIT.[6] The haemoglobin E, is the mostly prevalent in south east Asia. There is a double heterozygosity phenotyping in Haemoglobin E.[7]

Method

The study aimed to govern the prevalence and classify haemoglobinopathies and thalassemia in a major tertiary care center of capital province Punjab. The study also aims to highlight the commonest variants of Hb anormality. A proper preventive planning can be adopted if the screening of blood disorders is maintained.

Material and Methods

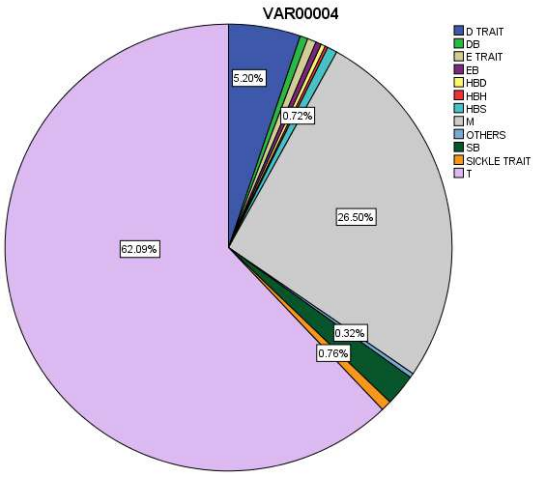
The study was based on a cross sectional survey technique. The study was conducted in a retro-spective domain.We selected a sample of around 12,000 patients for our study with purposive non-probability sampling. The sample size comprised of 12000 patients presenting with anaemia on a complete blood count. Out of 12000 samples collected, 357 samples were rejected because of clotted/hemolyzed samples, rest 11643 samples were included in the study. The duration selected for this study started from 1st January 2019 to 1st January 2020 (1 year study period). The venue for collection of blood samples was the institute of pathology, Chughtai lab, Lahore. The approval of Institutional review board was also sorted for this study. All the samples were collected from OPD and collection centers of the lab. The given set of blood samples were then undergone all the testing including CBC, sickling test, peripheral smear and Hb analysis. The complete blood count (CBC) was done using a Sysmex XN analzer, Gimsa staining were used in peripheral smears, the sebia analyzer helped in Hb electrophoresis. Percentages of different hemoglobin variants were noted.

Results

The sample number of 11643 blood samples were received for Hb electrophoresis. Out of these 11643 cases 9145 samples turned out to be normal. 2498(21.4%) cases were abnormal results on electrophoresis. The percentages of all the hemoglobinopathies were calculated. Out of 2498 cases 61.2% had beta thalassemia trait, 26.5% had beta thalassemia of type major, the HbD frequency trait was 5.2%, HbD homozygous 0.3%, sickle trait .8%, sickle cell disease 0.7%, HbE trait 0.6%, delta/beta thalassemia 0.6%, sickle/beta thalassemia 2.3%, HbE /beta thalassemia 0.4%, HbH disease 0.2%, and other rare cases constituted 0.3%. these rare cases included one case of homozygous HbE, one case od HbC/ beta thalassemia, 3 cases in bart zone, two cases in J zone and one case of sickle/delta thalassemia. The study revealed that (Beta) chain thalassemia was the most frequent hemoglobinopathy, HbD trait, the sickle beta thalassemia, sickle trait and sickle cell disease. Heterozygotes like delta/beta, HbE/ beta thalassemia were less common.

Table 1: *Frequencies of Different Haemoglobinopathies*

	Frequency number	Percent
D TRAIT	130	5.2
DB	14	.6
E TRAIT	16	.6
EB	10	.4
HBD	8	.3
HBH	5	.2
HBS	18	.7
M	662	26.5
OTHERS	8	.3
SB	57	2.3
SICKLE TRAIT	19	.8
T	1551	62.1
Total	2498	100.0



Pie Chart Showing Distribution Of Various Haemoglobinopathies

Discussion

A frequency of 21.4% was found relevant in our study for the given sample of patients, this is in a close comparison to a local study conducted by (waheed, 2012) on a sample of 504 patients showing frequency value of 28.4% . Another local study at Liaqat medical university showed the frequency of hemoglobinopathies at a value of 15% which is less than our study.[8] One more study conducted in Karachi showed 34% cases of hemoglobinopathies.[8] Another local research in the capital of province Khyber (KPK) showed higher value of haemoglobinopthies varying around 48.9% incontrast to 21.4% in our scenario.[9] An interesting finding in our research showed out of 2498 cases (21.4%) abnormal haemoglobinopathies the commonest was (61.2% beta thalassemia trait) and the second in line was (26.5% had beta thalassemia of type major). These results of commonest haemoglobinopathies are in real comparison with the international literature studies conducted in the region of Middle east, Central Africa, South Europe, Medieterranean region, and South Asia.[10,11] Apart from the two major haemoglobinopathies identified in our study the less common haemoglobinopathies included HbD trait was 5.2%, HbD homozygous 0.3%, sickle trait .8%, sickle cell disease 0.7%, HbE trait 0.6%, delta/beta thalassemia 0.6%, sickle/beta thalassemia 2.3%,HbE /beta thalassemia 0.4%, HbH disease 0.2%, and other rare cases constituted 0.3%. these rare cases included one case of homozygous HbE, one case of HbC/ beta thalassemia, 3 cases in bart zone, two cases in J zone and one case of sickle/ delta thalassemia. The study conducted in East Asia (KOREA) showed sickle cell trait to be the major haemoglobinopathies in contrast to our value of 0.8%.[18] Our study similarly showed prevalence of Hb Sickle trait on a very low value of 0.8% which is in contrast to the HbS 25% mounting in region of Middle Eastern population.[12] However our research stands in close co-ordination with the values of the most common and the least common haemoglobinopathies. Commonest haemoglobinopathy Beta thalassemia trait and HbS (SICKLE) being the least common haemoglobinopathy is in comparison with the international distribution of haemoglobinopathies.[13] Iron deficiency is common in major population of Pakistan, which might add as a confounding factor in the diagnosis of Beta thalassemia trait. This limitation can be resolved by identification and treatment of Iron deficiency.[14,15,16]

Conclusion

Thalassemia and hemoglobinopathies are serious health problems worldwide. Adequate measures should be taken for premarital screening for the detection of carriers. An appropriate screening plan should be introduced at prenatal level so that the haemoglobinopathies (disorder in HB) can be reduced. The screening would help in the healthy wellbeing of the children, their mental health and also their physical wellbeing. A healthy child brings along less of social financial burden on the effected family. Prevention is better than cure. And so does screening help in the morbidity of the situation. A regular policy has to be adopted for screening before marriage. Families with a history of thalassemia should also be targeted and counselling seminars should be conducted to bring awareness among general public.

Conflict of Interest: The study bears no conflict regarding interests.

Study Funding Source: Not required

Study Ethical Approval: Given

Authors Contribution: The authors contributed equally in accordance with ICMJE guidelines.

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