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Original Research Paper

Pathology

EPIDEMIOLOGICAL PROFILE AND PERIPHERAL BLOOD SMEAR ANALYSES OF THALASSEMIA PATIENTS AT TERTIARY HEALTH CARE CENTRE, RAJASTHAN

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ABSTRACT

Background: Thalassemia is an inherited disorder of haemoglobin production resulting from unbalanced haemoglobin synthesis. Thalassemia is predominantly a disease of children.

Material and Methods : This study was conducted on 100 patients of thalassemia major registered in PBM and associated group of hospitals,Bikaner in the years 2019 and 2020.Analysis of epidemiology and PBF abnormalities of the patients was done.

Results : It was found that maximum number of patients were in 0 to 5 years age group.Maximum frequency was in B positive age group,Hindu:Muslim ratio was 63:37 and male:female ratio was 54:46.PBF showed anisocytosis and poikilocytosis of RBC with microcytic hypochromic anemia,target cells and NRBCs as the main anomalies.White cells and platelets were normal in majority of cases.

Conclusion: Knowledge of epidemiological profile aids in implementation of screening and control programmes. PBS examination is an early,cost-effective investigation in the initial diagnosis of thalassemia.

KEYWORDS : Thalassemia, Epidemiology, PBS:Peripheral Blood Smear, PBF:Peripheral Blood Film, NRBC:Nucleated Red Blood Cells

INTRODUCTION

Thalassemia is a congenital anaemia due to deficient synthesis of one or more of the globin subunits of the normal human haemoglobin. The primary defect is usually quantitative, consisting of the reduced or absent synthesis of normal globin chains, but there are mutations resulting in structural variants produced at reduced rates and mutations producing hyper unstable hemoglobin variants with a thalassemia phenotype.(1)

This is a genetically transmitted condition. It is an autosomal recessive disorder.Thalassemia is a major health problem, placing an immeasurable emotional, psychological and economic burden on millions of people around the world.

The classification of thalassemia is based on the type of globin chain that is deficiently synthesized or the clinical expression of the disease.

Classification according to the type of globin chain which is deficiently synthesized **:** The two most common types are α (alpha) and β (beta) thalassemias. Less common types are δβ (delta beta) thalassemia and γδβ (gamma-delta-beta) thalassemia.

Classification according to clinical severity :Thalassemias have been clinically classified on the basis of severity of anaemia into three types—thalassemia major, thalassemia intermedia and thalassemia minor. Patients with severe transfusion dependent anaemia are said to have thalassemia major. In thalassemia minor, affected individuals are usually asymptomatic with mild or no anaemia in spite of prominent red cell abnormalities in peripheral blood. Thalassemia intermedia is of intermediate clinical severity.

The clinical syndromes associated with thalassemia arise from the combined consequences of inadequate haemoglobin production and imbalanced accumulation of globin subunits. The former causes hypochromia and microcytosis; the latter leads to ineffective erythropoiesis and hemolytic anemia(2)

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MATERIAL AND METHODS

The current study is a hospital based prospective analysis conducted at the Department of Pathology, Sardar Patel Medical College, Bikaner. 100 thalassemia major patients registered in P.B.M & associated group of hospitals in 2019 and 2020 were recruited for the study.Epidemiological profile of patients was noted and PBS of the patients were analysed after staining with Leishman's stain.

RESULTS

After thoroughly analysing the patients, all findings were studied and recorded to reach the following observations:

Table 1: Depicting age-wise distribution of cases

S.No.	Age Group	Percentage of Cases
1	0-5 Years	42
2	6-10 Years	33
3	11-15 Years	13
4	16-20 Years	09
5	21-25 Years	03
	Total	100

The table shows that maximum number of patients were from 0-5 years age group (42%),followed by 33% in the 6-10 years age group.

Table 2: Mean value of age

	Mean	Standard Deviation	Maximum	Minimum
Age (years)	8.14	5.297	22	1

The mean value of age was found to be 8.14±5.30 years, as illustrated in the above table.The maximum age in the study group was 22 years.

Table 3: Distribution of patients in different blood groups

Blood Group	Frequency	Percentage
A +ve	28	28
A –ve	1	1
B +ve	31	31
B –ve	2	2
O +ve	27	27
O –ve	5	5
AB +ve	6	6
Total	**100**	**100**

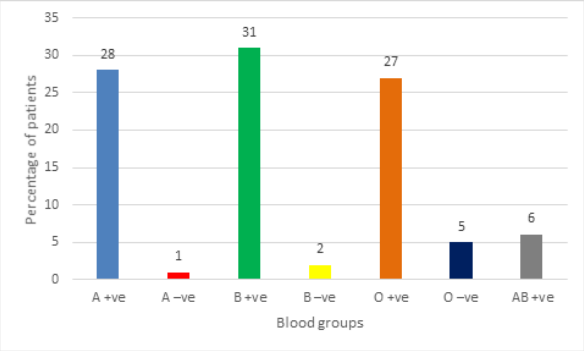


Figure 1: Distribution of patients in different blood groups

Figure 1 shows that majority of the patients were from B+ve followed by A+ve and O+ve blood groups with 31%, 28% and 27% prevalence respectively, as illustrated above in table 3.

Table 4: Gender-wise distribution of the study group

Gender	Frequency	Percentage
Male	54	54
Female	46	46
Total	**100**	**100**

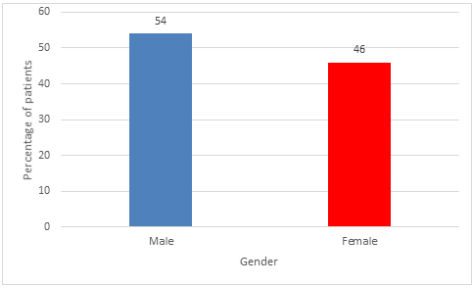


Figure 2: Gender -wise distribution of the study group

Table 4 indicates that 54% of patients were male and 46% were female as depicted in figure 2.

Table 5 : Religion-wise distribution of the study group.

Religion	Frequeny	Percentage
Hindu	63	63
Muslim	37	37
Others	00	00
Total	**100**	**100**

The table number 5 shows that Hindu:Muslim ratio was 63:37 in the study group as depicted below in figure number 3.

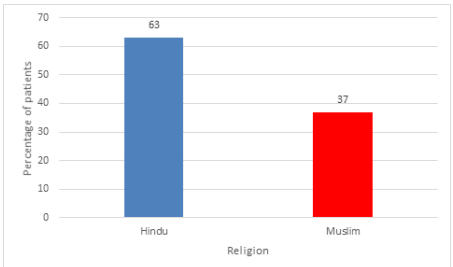


Figure 3: Religion-wise distribution of the study group.

Table 6 Table showing residential distribution of studied cases

Residence	Percentage of cases
Bikaner	66
Kolayat	7
Nokha	5
Pugal	4
Churu	3
Shri Dungargarh	3
Phalodi	3
Lunkaranshar	2
Napasar	2
Sujangarh	2
Bap	1
Gharsana	1
Khajuwala	1
Total	100

Table number 6 depicts that maximum number of cases were from Bikaner city(66%) followed by 7% from Kolayat.

Table 7: Prevalence of different red blood cell (RBC) types in the PBF

Size of RBC	Frequency	Percentage
Micro+ Normo	58	58
Micro	42	42
Total	**100**	**100**

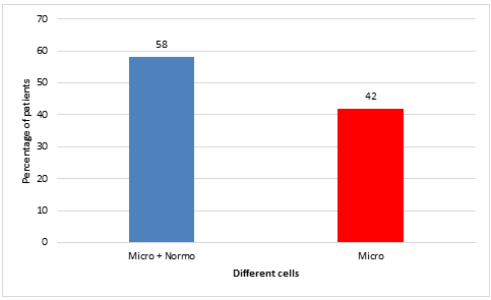


Figure 4: Prevalence of different red blood cells in the study group.

In the study group, it was observed that 58% patients had mixed population of microcytic and normocytic cells(micro+normo), while 42% had microcytic (micro) cells, as illustrated in Table 7 and Figure 4.

Table 8: Degree of anisocytosis of Red Blood Cells (RBC)

Anisocytosis	Frequency	Percentage
Severe	14	14
Moderate	43	43
Mild	43	43
Total	**100**	**100**

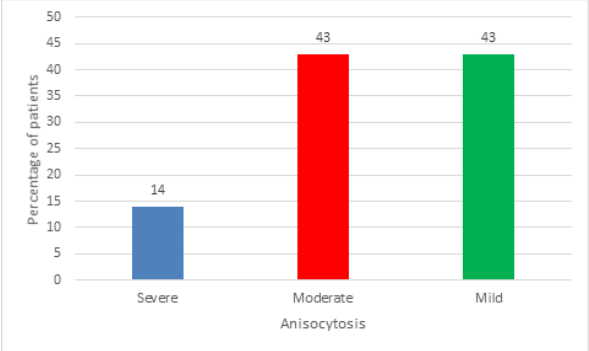


Figure 5: Distribution of anisocytosis of RBC

All the patients had anisocytosis,that is,variation in size of RBCS.14% of the patients had severe,43% had moderate and 43% had mild anisocytosis.

Table 9: Change in Total Leukocyte Count (TLC)

TLC	Frequency	Percentage
Normal range	91	91
Raised	9	9
Total	**100**	**100**

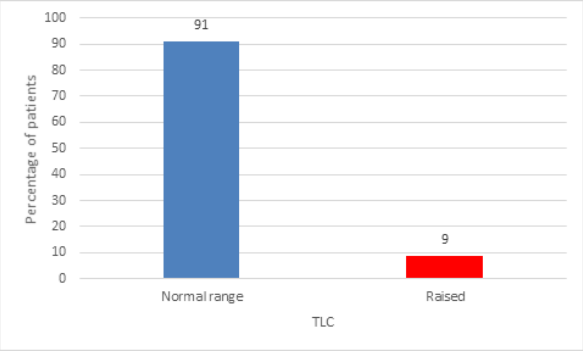


Figure 6: Depicting change in TLC

As depicted above, raised TLC was found in 9% patients while the TLC was within normal range in 91 % of patients.

Table 10: Change in platelet count

Platelet Count	Frequency	Percentage
Normal range	85	85
Raised	2	2
Reduced	13	13
Total	**100**	**100**

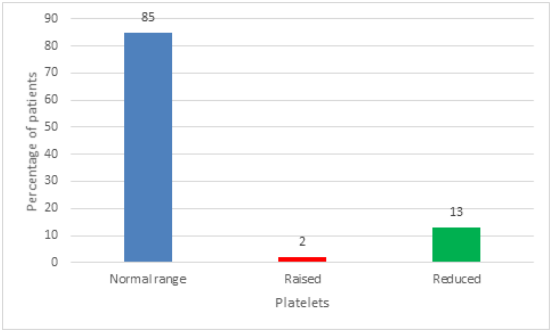
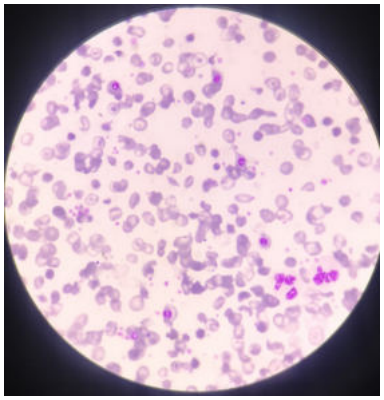


Figure 7: Change in platelet count

As illustrated in Table 10 and Figure 7, platelets were raised in only 2% patients, reduced in 13% patients,while 85% patients had platelet count within the normal range.



Picture of PBS depicting microcytic hypochromic RBCs along with target cells, tear drop cells and nucleated RBC.

The mean value of NRBCs was 5.35/100WBCs.The WBCs and platelets were in the normal range in majority of thalassemia patients.

DISCUSSION

Thalassemia is one of the most common of hemoglobinopathies. Thalassemias are inherited defects in globin chain synthesis.These disorders are the result of the existence of an abnormal allele in one or more globin genes. The decrease or loss of an or chain has unfavourable effects on the production and the survival of red blood cells. This may cause a decrease in the concentration of the globin chain and hemoglobin, resulting in microcytosis and hypochromia. Iron deficiency anemia and thalassemia are recognized as the most important causes of hypochromia and microcytosis(3,4).

There are more than 200 known thalassemia mutations, the majority being single nucleotide substitutions, insertions or short deletions. Few large gene deletions have been identified and most of them can be diagnosed by Gap–PCR.(5)

Screening can be targeted at different age groups.Numerous carrier screening programmes are conducted world-over at the premarital or early pregnancy levels.Recently,in some Islamic high-risk countries such as Iran,Saudi Arabia and Palestinian territories,hemoglobinopathy screening has become mandatory for all couples before taking the approval to get married.(6,7)

In our study, 58% patients had microcytic and normocytic RBC, while 42% patients had microcytic cells only. Lower prevalence of microcytic anemia were reported by Mishra *et al* in 2018 (8) and Das *et al* in 2020. (9)

In the present study, on analyzing the degree of anisocytosis amongst the study group,severe anisocytosis was found in 14% of the patients only, while 43% patients had moderate and 43% had mild anisocytosis.

England and Fraser in 1979 (10) after evaluating 1500 peripheral smears in β thalassemia cases, found microcytic hypochromic red cells and significant target cells in RBC series. In another study,Santhi in 2010 (11) showed mild to moderate microcytic hypochromic red cells with mild anisocytosis and 5% of cases showed target cells .In our study,68% of patients showed target cells and the mean NRBC value was 5.35 /100 WBC in the PBS.Tear drop cells and few schistocytes were the other RBC abnormalities.

CONCLUSION

Thalassemia was found to be more prevalent in childhood .The mean value of age in the study group was 8.14±5.30 years.Majority of patients had B+ve blood group(31%) followed by A+ve (28% patients) and O+ve (27% patients) blood groups. Males were more often affected as compared to females . The Hindu: Muslim ratio was found to be 63:37 and there was no patient from other religions.

PBS assessment revealed microcytic hypochromic anemia with anisopoikilocytosis of RBCs .Mild or moderate anisocytosis was commonly observed with severe anisocytosis seen in only 14% of cases.Target cells, tear drop cells and NRBC were found to be the predominant RBC anomalies in the PBS.There was no significant alteration in WBC and platelets. This study highlights the importance of PBF examination in diagnosis of thalassemia.

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