ORIGINAL RESEARCH PAPER
Volume - 13 | Issue - 07 | July - 2024 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

STUDY ON CLINICOPATHOLOGICAL PROFILE OF MULTITRANSFUSED THALASSEMIC PATIENTS

Pathology

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ABSTRACT

Background & Aims: Thalassemia's are inherited blood disorders characterized by decreased hemoglobin production. If adequate transfusion is possible affected children grow and develop normally and have minimum abnormal physical sign. The disease poses a great problem due to the effects of iron loading resulting from ineffective erythropoiesis and repeated blood transfusion. This study is meant to find out effect of multiple blood transfusion in thalassemia patients on growth and evaluation of hepatic and thyroid function along with effect of iron on reticuloendothelial system. **Material & Methods:** This longitudinal study was conducted on 100 beta thalassemia major patients at the Department of Pathology over a period of two years. The present study will be conducted for detection of clinical and pathological profile of multi transfused thalassemic patients i.e. complete blood count (CBC), Peripheral smear examination findings, HPLC findings, HbA1C value, thyroid and liver function test, Serum ferritin and correlation between them. **Results:** All patients had markedly elevated serum ferritin levels indicating iron overload. We observed that as the serum ferritin increases HbA1C value increases. Frequent blood transfusions can control hemoglobin level, but if serum ferritin levels are greater than desired level, patient's physical growth can be affected. **Conclusion:** Frequent monitoring and observation of the multitransfused thalassemic patient has an immense importance to prevent patient from going into severe organ impairment. Judicious use of serum ferritin in multi transfused thalassemic major patients is of prime importance to prognosticate disease severity.

KEYWORDS

Thalassemia, Multitransfused patient, Serum ferritin level.

INTRODUCTION:

Thalassemia's are inherited blood disorders characterized by decreased hemoglobin production[1].

Thalassemia is a recessively inherited heterogenous group of disorder. It results from inability of the red cells to synthesize an adequate amount of either a or ß chain in the hemoglobin molecule[2].

Anemia typically appears within the first few months of life and gets worse over time. If adequate transfusion is possible, affected children grow and develop normally and have minimum abnormal physical sign. The disease poses a great problem due to the effects of iron loading resulting from ineffective erythropoiesis and repeated blood transfusion. Inadequately transfused child develops typical features of Cooley's anemia. Stunted growth with bossing of skull, overgrowth of maxillary region and face gradually assuming mongoloid appearance[3].

Patients who get hypertransfusion treatment, which keeps the hemoglobin level at a minimum baseline level [12 gm%], had longer life expectancies and improved quality of life[4]. First indication of iron overload is usually the absence of pubertal growth spurt and failure of menarche. In Indian children the main problem is failure to maintain adequate hemoglobin level due to inadequate transfusion. Additionally, the patients cannot afford or access to facilities for regular chelation therapy, which removes excess iron. Hyperpigmentation, hepatic fibrosis and cirrhosis, growth failure, delayed puberty, and endocrinopathies such as diabetes mellitus, hypogonadism, hypoparathyroidism, and hypothyroidism are all detrimental effects of transfusional hemosiderosis[5].

In second decade the patients face the consequence of myocardial hemosiderosis, the leading cause of death in these patients. Iron deposition had been implicated as a cause of hypothyroidism, which is mainly subclinical in nature[1,6].

AIMS AND OBJECTIVES:

- Study of iron overload in patients having repeated packed cell transfusion by estimation of serum ferritin in multi transfused thalassemic patients.
- Bio chemical evaluation of liver & thyroid function and blood sugar with HbA1C detected by HPLC.
- Analysis of various laboratory aspects namely hematological parameters, peripheral blood smear examination and HPLC findings.
- Clinical examination of thalassemia patients to find out effect of disease on their growth i.e. Height, Weight, Organomegaly etc.

MATERIAL AND METHOD:

Design Of Study: longitudinal study

Duration Of Study: 2 years study

Sample Size: Total 100 cases

Study Area: Department of pathology.

Patients who are known case of beta thalassemia major and received regular packed cell transfusion were included in the study. Patients having multiple congenital anomalies along with thalassemia, Hemoglobinopathies and Chronic hemolytic anemia other than thalassemia were excluded from the study.

The present study will be conducted for detection of clinical and pathological profile of multitransfused thalassemic patients i.e. complete blood count (CBC), Peripheral smear examination findings, HPLC findings, HbA1C value, thyroid and liver function test and it's correlation with HbA1C and serum ferritin.

RESULTS

In present study, most patients were from 16-20 year age group and male predominance.

It was observed that a higher frequency of transfusion (twice per month) was seen in the older age group and lower frequency (once per month) was seen in the younger age group.

Frequency of multitransfused thalassemic patients according to age at time of diagnosis, 72 cases were diagnosed at age of 1 year. Only 11 cases were diagnosed after 2 years of age. However exceptionally 2 case were diagnosed at 6 years and 8 years of age.

MCV and MCH were found to be low normal values (in case of MCV) and moderately reduce (in case of MCH) were found to be dependent on transfusion frequency or ferritin values.

Correlation factors of various parameters are as follows,

RBC mass : 0.2267
Hb(gm%) : 1.865
MCH(pg) : 1.531
MCV(fl): 6.539

In Present study, we discovered that 10 patients had severe degree anemia, 54 patients had moderate degree anemia and 36 patients had mild degree anemia.

Table I : Severity Of Anemia Based On Hemoglobin Concentration Of Multitransfused Thalassemic Patients

Sr.No.	Hemoglobin	Total number of Cases	Percentage
1	<7 gm/dl	10	10
2	7 gm/dl to 10 gm/dl	54	54
3	>10 gm/dl	36	36
	Total	100	100

Peripheral Blood Film Examination:
Patients with history of recent blood transfusion show normocytic normochromic blood picture along with microcytic hypochromic blood picture and occasional target cells and elliptocytes.

HPLC Findings:
Table II : HbF Value In Multitransfused Thalassemic Patients

Sr No.	HbF value	Number of cases	Percentage
1	<2%	52	53
2	2% - 30%	45	45
3	>30%	2	2
	Total	100	100

Height And Weight:
According to the IAP growth chart, 66 cases were below the 3rd percentile in height, 26 cases were between the 3rd and 50th percentile, and only 8 cases were above the 50th percentile. 54 cases were below the third percentile in weight, 36 cases were between the third and 50th percentile, and only 10 cases were above the 50th percentile.

Organomegaly And Splenectomy Status :
Out of 100 subjects, 15 had variable degree hepatomegaly, 23 had splenomegaly, and 29 had both. There were 33 cases with no organomegaly. 01 case is known to had undergone splenectomy.

Serum Ferritin: All patients had markedly elevated serum ferritin levels indicating iron overload.

HbA1C by HPLC: 35% was pre-diabetic and 48% was diabetic. The serum ferritin increases, HbA1C value increases.

Thyroid Function Test: Thalassemia patients had been divided according to thyroid function into four groups.
Group-1: 56 cases –Normal thyroid function. Normal T4 & TSH level.
Group-2: 27 cases - Compensated hypothyroidism. Normal T4 & ↑ TSH level.
Group-3A: 09 cases - Decompensated hypothyroidism. ↓ T4 & increased TSH level.
Group-3B: 08 cases - Patients having decompensated hypothyroidism with Pituitary dysfunction.

Serum thyroxine reduced but TSH normal/reduced. Normally reduced thyroxine level is accompanied by elevated TSH level.

All group does not have statistical significant difference in mean serum ferritin level.

Liver Function Tests: -
Serum Bilirubin Level: We have observed that 59 patients have elevated bilirubin level, 67 patients have elevated SGPT level. There is no significant difference in mean serum ferritin in patient having elevated and normal SGPT level.

Serum Albumin Level: We have observed that 11 patients have decreased serum albumin level.

DISCUSSION:

Thalassemia patients are virtually dependent on regular packed cell transfusion for their survival. Transfusion appears to be a double edged sword having its benefit as well as side effects.

Side effects primarily due to iron overload which in itself is quite cumbersome.

In present study, Peripheral blood smear examination show no effect with the number of transfusions, average serum ferritin level throughout the year, age and sex.

We have observed that HbF is protective for thalassemic patients which leads to less iron overload in multitransfused thalassemic patients.

Iron overload occurs very rapidly in patients who are on chronic transfusion. Patients who are being transfused every three or four week gain 0.5 mg/kg per day of iron in excess of natural losses. Retardation of both height & weight seen in thalassemia patients. As per IAP growth chart, in our study 66 cases were stunted(<3rd percentile) for their height. The proportion of short stature in our study was similar to the previous studies of Vyas kumar et al., Jana et al.,Fadlayana et al., and ParthaSarathiDas et el., which discovered the incidence of short stature 65.71%, 65.8%, 62% and 67% respectively. The cause of short stature in thalassemic children can be chronic anemia, hypersplenism and folate deficiency. Madeddue et al[7] showed presence of growth retardation in their study in several patients in their study during first year of life and the retardation affected all the thalassemia patients when they reached puberty.

The concentration of ferritin is directly proportional to the total iron stores in the body, resulting in serum ferritin concentrations becoming a common diagnostic tool in the evaluation of iron status[8]. In present study, Serum ferritin level was elevated in all the patients of beta thalassemia major. Iron overload initially leads to glucose abnormalities such as insulin resistance and hyperinsulinemia, which is followed by diabetes mellitus. This type of diabetes was called bronze diabetes due to the greyish colour of skin developed from the deposition of excess iron.

Abnormal liver function appears to be related to the high serum ferritin levels. In present study, no children had any symptom or sign related with thyroid dysfunction. This hypothyroidism often goes undetected because less attention is paid to it. Agarwal et al[9] found 20% incidence of hypothyroidism in their study. Madeddu et al, Spitz IM et al[10] also observed hypothyroidism with ↓ T4 & ↑ TSH. From this discussion it is evident that hypothyroidism is present in a significant number of transfusion dependent thalassemia patients. Iron overload had been implicated as an etiological factor for hypothyroidism. From this discussion it is evident that regular transfusion leads to deposition of iron in various tissue so that regular evaluation of thyroid function, hepatic function and level of HbA1C is mandatory in multitransfused thalassemic patients.

CONCLUSION:

Judicious use of serum ferritin in multi transfused thalassemic major patients is of prime importance to prognosticate disease severity. Liver function tests showed alterations which includes increase in bilirubin and SGPT level and lower level of albumin. Thyroid function tests showed subclinical hypothyroidism in these patients. Alteration in liver and thyroid function tests shows the effect of iron overload on the organs. So, frequent monitoring and observation of the multitransfused thalassemic patient has an immense importance to prevent patient from going into severe organ impairment.

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