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Original Research Article

Descriptive Observational Assessment of the Clinico-Pathological Profile of Pediatric Patients with Thalassemia Major

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Abstract

Aim: To study the clinical profile and laboratory parameters of Thalassemia major patients between the age group 1-14 years.

Material & Methods: It was a Descriptive- Observational study carried out in the Department of Pediatrics & Neonatology, RIMS, Ranchi, Jharkhand, over a period of one year. This study was conducted on 160 children with β-Thalassemia major patients aged between 1-14 year.

Results: Serum Phosphorus values with majority (68) of children showed decreased level. Majority of participants belongs to 1-5 years (N=48) which shows statistical significance (0.0002).

Conclusions: Thalassemia needs greater public awareness and prevention strategies in our country. Some communities are at high risk as compared to others. Education programs and compulsory antenatal screening appear to be the order of the day.

Keywords: Beta Thalassemia, Congenital Hemolytic Anemia, Hb E Beta Thalassemia.

Introduction

Haemoglobinopathy and thalassaemia constitute a major bulk of congenital hemolytic anemia in India. They cause a high degree of morbidity, moderate to severe haemolytic anaemia among infants and children and several deaths in India. [1] Congenital hemolytic anemia is anemia due to hemolysis, the abnormal breakdown of red blood cells either intravascular or extravascular. Anemia results If the rate of destruction exceeds the capacity of the marrow to produce red blood cells. Inherited RBC defects of structure and metabolism may result in a chronic hemolytic state, that includes - hemoglobinopathy, like sickle cell anaemia, α thalassemia, β thalassemia, HbE β-thalassemia; RBC enzyme defect, like glucose 6 phosphate dehydrogenase deficiency; RBC membrane disorders like hereditary spherocytosis.[2] Haemoglobinopathies affect 4.5% of the world population.[3]

The prevalence of β- thalassemia trait varies between 3-17% because of consanguinity and caste and area endogamy.[4]Every year, ten thousand children with β-thalassemia major are born in India, which constitutes 10% of the total number in the world.[5]HbE thalassemia is common in north-east parts of India.[6]

Beta thalassemia is the commonest inherited hemoglobin disorder in the Indian subcontinent with an uneven distribution among the different endogenous populations. Carrier frequency ranges between 3.7 and 10 % [7]. With national programmes the epidemiology of the disease has changed worldwide. However, in India, though immense progress has been made in the management and antenatal diagnosis of the disease, an accurate estimation of the disease burden at the district level is as yet not available [7, 8].

Endocrine functions of Thalassemic children progressively decline with age secondary to hemosiderosis and nutritional deficiencies, resulting in multiple endocrinopathies like hypothyroidism, growth hormone deficiency, delayed puberty, diabetes mellitus and osteoporosis. So, all these children require regular monitoring of endocrine functions after 5 years of age, and regular monitoring of height, weight semi-annually.

Thus, we aim to study the clinical profile and laboratory parameters of Thalassemia major patients between the age group 1-14 years.

Material & Methods:

It was a Descriptive-observational study carried out in the Department of Pediatrics & Neonatology, RIMS, Ranchi, Jharkhand over a period of one year. This study was conducted on 100 children with β-Thalassemia major patients aged 1-14 year.

Inclusion criteria:

All the diagnosed β-Thalassemia major patients between the age group of 1 to 14 years attending the Department of Pediatrics & Neonatology, RIMS, Ranchi, Jharkhand.

Exclusion criteria:

1. Patients are less than one year and more than 14 years of age group.
2. Children having multiple congenital anomalies along with Thalassemia major.
3. Coexisting cardiac or pulmonary disease.
4. Chronic haemolytic anaemia, other than β- Thalassemia major.
5. Thalassemia minor
6. Beta –thalassemia with any other haemolytic anaemia

Methodology

A preformed and pre-checked proforma was used for data collection that included personal information, data regarding the number of transfusions and pre-transfusion haemoglobin and serum ferritin, at what dose of chelators they were with clinical examination finding and laboratory investigation reports.

Transfusion index is calculated by the formula blood volume received in ml/kg/month. The serum ferritin level was measured in all Thalassemic patients. Iron chelating agents were advised to all patients with serum ferritin level above 1000 ng/ml. Haemoglobin was measured before transfusion by Sahli's method. Blood group cross-matching was done by blood typing. Standard references were used. Data regarding various clinical and laboratory parameters were recorded and tabulated and presented as frequency, percentage and mean.

Statistical analysis

The data collected were analysed with Graph Ped statistical software version 7.0. Continuous variables were presented as mean for parametric data. The student t test was applied for the calculation of statistical significance whenever the data followed normative distribution.

Nominal categorical data between the groups have been compared using Chi-square test or Fisher's exact test as appropriate. $P < 0.05$ was taken to indicate a statistically significant difference.

Correlation analysis was done by calculating the correlation coefficient.

Results:

Basic demographic details were seen in table 1 with majority of study participants being males (79%) and 21% were females.

Table 3 shows Serum Phosphorus values with majority (68) of children showed decreased level.

Majority of study participants (70) showed an increased Serum Alkaline Phosphatase level of >8 as seen in table 4

Table 5 shows a statistically significant (>0.05) Correlation of liver function test, renal function test with serum ferritin.

Table 6 shows correlation of Hepatomegaly with transfusion index and different range of serum ferritin in all three groups. Majority of participants belongs to 1-5 years (N=48) which shows statistical significance (0.0002)

Table 1: Distribution of cases according to sex.

Sex	No of cases (N=100)
Females	79
Males	21

Table 2: Showing distribution of patients according to Height for age (n=100).

	Short stature (below 3rd centile)	Normal stature
Male	19	37
Female	14	30
Total	33	67

Table 3: Serum Phosphorus values (mg/dl).

Age (Years)	Decreased	Normal	Increased	Total
1-3	13	1	9	23
4-11	33	14	21	68
12-14	1	2	6	9
Total	47	17	36	100

Table 4: Serum Alkaline Phosphatase (I/U).

Age (Years)	Normal	Increased	Total
<8	45	25	70
>8	6	24	30
Total	51	49	100

Table 5: Correlation of liver function test, renal function test with serum ferritin.

Dependent Indices	Correlation to Serum ferritin (R square)	P value
SGOT	0.0131	p>0.05
SGPT	0.0251	p>0.05
Serum Bilirubin Total	0.0155	p>0.05
Serum Bilirubin Direct	0.00162	p>0.05
Total Serum Protein	0.0177	p>0.05
Serum Albumin	0.0128	p>0.05
Serum Urea	0.0112	p>0.05
Serum Creatinine	0.00671	p>0.05

Table 6: Showing correlation of Hepatomegaly with transfusion index and different range of serum ferritin in all three groups.

Age Group	S. Ferritin	Transfusion Index
1-5 years (N=70)	**<500-1000 (n=4/10) 7.62±4.4 cm**	**r = -0.478 p=0.0002 (44.2 ±191.8)**
	1001-1500 (n=13/21) 8.98±2.62 cm	**r = - 0.571 p = 0.571 (11.24±21.3)**
	1501-2000 (n=6/7) 8.61±2.32 cm	**r =0.261 p=0.477 (15.72±6.27)**
	>2000 (n=10/12) 8.81±1.38 cm	**r = - 0.521 p = 0.271 (14.2±9.81)**
6-10 years (N=65)	**<500-1000 (n=7/9) 8.22±2.71 cm**	**r = - 0.611 p = 0.132 (54.5±94.5)**
	1001-1500 (n=3/3) 9.59±1.63 cm	**r = - 0.621 p = 0.691 (20.2±4.2)**
	1501-2000 (n=3/3) 110.2±4.71 cm	**r = - 0.082 p = 0.776 (152.1±212.2)**
	>2000 (n=16/18) 11.2±2.27cm	**r = - 0.271 p = 0.467 (41.2±70.87)**

Discussion:

Thalassemia can be explained by the fact that these cases having a milder clinical course and thus presenting at a later age compared to other group of patients of congenital hemolytic anemia, live longer and also get the opportunity to come under medical attention. In our study there was more prevalence of congenital hemolytic

anemia in males in respect to females (63 % versus 37%). However it is difficult to conclude as the study sample is small. In this study, the mean hemoglobin was found to be lowest in patients of β thalassemia (5.1gm/gl) and HbE β thalassemia (5.8 gm/dl). The mean total serum bilirubin was found to be highest among β Thalassemia patients (3.0mg/dl). Our results are comparable to previous studies. [9-12]There were a wide spectrum of clinical manifestations among patients of congenital hemolytic anemia.

Serum urea was high in 65(64.35%) cases [27-28]. Though most of the urea and creatinine were only mildly elevated which may be due to chelator therapy in the higher age group.

Contributing factors to growth retardation include recurrent infections, nutritional deficiency, chronic hypoxia, iron toxicity from transfusion hemosiderosis, poor transfusional status and inadequate chelation [13-14]. Hemolytic facies were present in 39 % of patients and 78 % came with moderate to severe anemia. This gives an insight to late presentation in our country as compared to the west where the mean Hb at diagnosis has been reported as 8.2 gm/dl [15].

The severe and most frequently encountered hemoglobinopathies in India include the thalassemia and sickle cell disease. These hemoglobinopathies are geographically distributed in the country. TM is the commonest hemoglobinopathy seen in the northern part of the country, sickle cell being confined mainly to central India [7-8].

Kattamis et al. [16] in 1975 noted age at presentation to be 13.1 (2–36) months. Cao [17] reported the mean age of children who presented with TM to be 8.4 ± 9.1 months. In 1984, Modell and Berdukas [18] reported 60 % of patients to present in the first year of life, mean age being 6 months.

Conclusion:

Thalassemia needs greater public awareness and prevention strategies in our country. Some communities are at high risk as compared to others. Education programs and compulsory antenatal screening appear to be the order of the day.

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