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ORIGINAL RESEARCH

Clinical profile of children with thalassemia admitted at a tertiary care hospital

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ABSTRACT
Thalassemia syndromes are heterogeneous because of the various possible mutations which affect the human globin chain loci, such as globin chain initiation, translation, termination and also gene deletion. All enrolled children were taken detailed clinical history including age at first blood transfusion, number of blood transfusion, duration of iron chelation therapy and General physical examination findings were recorded on a predesigned proforma. Before blood transfusion, for all enrolled children one blood sample was sent for Serum ferritin level and another sample for pre-transfusion Hb. Serum ferritin level was measured by electrochemi-luminescence technique using Cobas 6000 analyser. In the study, all children (100%) presented with Easy Fatigability and progressive pallor. All children except 2 (4.4%) had presented with cough secondary to URTI. None of the children presented with rapid breathing, chest in drawing, Chest pain. On per abdomen examination all children had hepatomegaly (100%) and 34 children (75.5%) had splenomegaly, remaining 11(24.5%) children were splenectomised.

Key words: Clinical profile, children, thalassemia

INTRODUCTION

Thalassemia refers to genetic disorders in globin chain production. In individuals with beta thalassemia, there is either a complete absence of β globin production (β-thalassemia major) or a partial reduction in β globin production (β thalassemia minor). In alpha thalassemia, there is an absence of or partial reduction in α globin production.It was traditionally considered a paediatric disease as it presents within 1st year of life and in the past, majority of the patients died in adolescence[1].

Thalassemia syndromes are heterogeneous because of the various possible mutations which affect the human globin chain loci, such as globin chain initiation, translation, termination and also gene deletion.

These mutations caused decreased or absent β chain production leading to excessive α chains. These α chains become unstable and precipitate in red cell precursors, in the absence of β chains, producing intracellular inclusions which interfere with maturation of red cells. This leads to accelerated apoptosis and hence ineffective erythropoiesis.(Severity of the disease is determined by this imbalance between α and β globin chains rather than the decreased production of Haemoglobin) [2, 3].

Combination of this ineffective erythropoiesis, shortened red cell survival due to α globin inclusions and progressive splenomegaly is responsible for the anaemia in β thalassemia. A stress situation is produced in the bone marrow which in turn stimulates erythropoietin synthesis causing bone marrow expansion, leading to serious skeletal deformities[4].

β Thalassemia major (also called Cooley's anaemia) usually manifests during the first year of life, once the switch from the production of fetal Hb(α2γ2) to that of adult Hb (α2β2) occurs.Affected children fail to develop normally and their growth is retarded from shortly after birth. They are sustained only by repeated blood transfusion, which improve the

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anaemia and reduce the skeletal deformities associated with excessive erythropoiesis. With transfusion alone survival into second or third decade is possible, but gradually iron overload develops. The combination of iron present in transfused red cells and gut lead inevitably to iron overload[5, 6].

METHODOLOGY

STUDY TYPE: Hospital based cross sectional study.

STUDY POPULATION: Children aged between 5-15 years with beta thalassemia major admitted for periodic blood transfusion in paediatric ward.

INCLUSION CRITERIA

Children with confirmed diagnosis of beta thalassemia major in the age group 5 years to 15 years.

EXCLUSION CRITERIA

1. Thalassemiachildrenwhowerealreadydiagnosedcasesofpulmonary dysfunctions. (i.e. asthma, bronchiectasis and other chronic lung diseases).
2. Children with congenital heart disease/Rheumatic heart disease.

- Ethical Clearance was obtained from the Institutional Ethics Committee.
- Children who fulfill the inclusion/exclusion criteria for the study were selected.
- Informed and written consent was obtained from parents of all cases.
- All enrolled children were taken detailed clinical history including age at first blood transfusion, number of blood transfusion, duration of iron chelation therapy and General physical examination findings were recorded on a predesigned proforma.
- Before blood transfusion, for all enrolled children one blood sample was sent for Serum ferritin level and another sample for pre-transfusion Hb.
- Serum ferritin level was measured by electrochemi-luminescence technique using Cobas 6000 analyser.
- Pulmonary function test was done using spirometer (RMS Helios) (annexure1), within 24hrs of blood transfusion.

RESULTS

Table 1: Age Distribution of Study Subjects (N=45)

Age	Number of Cases (N=45)
<10 years	37(82.2%)
≥10 years	8(17.8%)

Age of the children included in the study ranged from 5 year to 15 years. Thirty seven (82.2%) children were less than 10 years and eight children (17.8%) were more than or equal to 10 years. Median age was 7 years (Range: 5years-13years).

Table 2: Sex Distribution of Study Subjects (N=45)

Sex	Number of Cases (N=45)
Male	28(62.2%)
Female	17(37%)

62.2% were males and 37.8% were females with M:F ratio was 1.6:1.

Table 3: Baseline Charecteristics of Study Subjects (N=45)

Baseline Characteristic			Mean ± SD
Anthropometry	Weight (kg)		17.8±4.8
	Height (cms)		111.8 ± 13.7
	BMI		14.0 ± 1.4
No. of Blood Transfusion	<50 (N=9)		36.4 ± 6.9
	50-100 (N=19)		78.8±10.6
	>100(N=3)		106.3±3.0
Duration of Chelation (Years)	Total (N=45)		4.2±1.4
	< 5Years	N=25 (55.5%)	
	>5Years	N=20 (44.4%)	
Pre-Transfusion HB(gm/dl)			5.7±1
Post-Transfusion HB(gm/dl)			9.7±0.8
Serum Ferritin(ng/ml)	Total		3085.4 ± 2184.7
	<2500 (N=23)		1545±461.5
	>2500(N=22)		4695.6±2112.6

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Mean age was 7.78±2.4, mean height was 111.8±13.7 cms, mean BMI was 14±1.4, mean No of blood Transfusions were 62.1 ± 24.7, mean pre transfusion Hb was 5.7 ± 1.0%,mean post-transfusion Hb was 9.7±0.8and mean Serum Ferritin was 3085.4 ± 2184.7.

Table 4: Clinical Features among Study Subjects (N=45)

Clinical Features		Number of Cases (N=45)
Progressive Pallor		45(100%)
Easy Fatigability		45(100%)
Cough		2(4.4%)
Rapid Breathing		0
Chest in Drawing		0
Chest Pain		0
Hepatomegaly		100(100%)
Spleen	Splenomegaly	34(75.5%)
	Splenectomised	11(24.5%)

In the study, all children (100%) presented with Easy Fatigability and progressive pallor. All children except 2 (4.4%) had presented with cough secondary to URTI. None of the children presented with rapid breathing, chest in drawing, Chest pain. On per abdomen examination all children had hepatomegaly (100%) and 34 children (75.5%) had splenomegaly, remaining 11(24.5%) children were splenectomised.

DISCUSSION

There are few studies on lung function in beta thalassemia children in India. Lung dysfunction is among the least studied complication in children with beta thalassemia major, probably due the lack of pulmonary symptoms presenting compared to cardiomyopathy or endocrine complication. Different types of pulmonary function abnormalities have been described in TM patients. However, the precise etiology of the pulmonary dysfunction remains unknown. Several mechanisms for the pulmonary dysfunction in thalassemia have been investigated, including iron overload, allergy, and correlation with transfusion, but none of these has provided a satisfactory explanation[7, 8].

Ours was a cross sectional descriptive, hospital based study. A total number of 45 children included in our study, majority of these children were <10years (37 children) as compared to children >10years (8children). Median age was 7 years (5-13years). The mean age was 7.78 ± 2.420 years. In many other studies mean age was >10 years, this could be probably because of age difference in inclusion criteria[9, 10].

Male children affected more than female children with M:F ratio 1.6:1. Similar male preponderance observed in other studies. But these studies have not been able to prove a causal relationship for male preponderance.

All children presented with easy fatigability and progressive pallor. No respiratory symptoms except cough(secondary to URTI) which was found in only two children(4.4%).

CONCLUSION

In the study, all children (100%) presented with Easy Fatigability and progressive pallor. All children except 2 (4.4%) had presented with cough secondary to URTI. None of the children presented with rapid breathing, chest in drawing, Chest pain. On per abdomen examination all children had hepatomegaly (100%) and 34 children (75.5%) had splenomegaly, remaining 11(24.5%) children were splenectomised.

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