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Original Research Article

Assessment of the Clinico-Epidemiological Profile of Thalassemia Patients in a Tertiary Care Hospital in Bihar Region

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Abstract

Aim: To assess the pattern, clinical presentations, complications, and management practices among thalassemia cases.

Material & Methods: This is a retrospective record-based cross-sectional study was conducted in the Department of Pediatrics, Jawaharlal Nehru Medical College and Hospital, Bhagalpur, Bihar, India. The secondary data of all confirmed cases of thalassemia were examined by the investigators.

Results: The mean age of cases was 6.2 years. The age at diagnosis ranged from 0.1 to 11 years. The majority of cases were under-fives 31 (38.75%) and were male. Bone deformities were reported in 17 (21.25%) cases, all of which were beta thalassemia major cases.

Conclusion: Hemoglobinopathies are the commonest hereditary disorders in India and pose a major health problem. The data on the prevalence of β-thalassemia's and other hemoglobinopathies in different caste/ethnic groups of India is scarce.

Keywords: Hemoglobinopathies, β-thalassemia, Iron Chelating Agents

Introduction

The March of Dimes Global Report on birth defects estimates that 7.9 million infants are born annually with a serious birth defect. Most of these (7.4 million) occur in the middle and low-income countries [1]. The hemoglobin disorders, sickle cell anemias and thalassemia's contribute significantly to this global toll. Approximately 7 % of the world's population is a carrier for hemoglobin disorders with 300,000–500,000 births every year with the severe heterozygous form of disease [2–4]. 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 % [5–10].

The prevalence of β- thalassemia trait varies between 3-17% because of consanguinity and caste and area endogamy. [11] Every year, ten thousand children with β-thalassemia major are born in India, which constitutes 10% of the total

number in the world. [12] HbE thalassemia is common in north-east parts of India. [13] The only forms of treatment available for thalassemia patients are regular blood transfusion, iron chelation therapy in an attempt to prevent iron overload and the judicious use of splenectomy in cases complicated by hypersplenism.

The curative treatment like bone marrow transplantation is costly and so prevention is the cost-effective strategy, which includes population screening, genetic counseling and prenatal diagnosis. [14]

Thus, we aim to assess the pattern, clinical presentations, complications, and management practices among thalassemia cases.

Material & Methods:

This is a retrospective record-based cross-sectional study was conducted in the Department of Pediatrics, Jawaharlal Nehru Medical College and Hospital, Bhagalpur, Bihar, India for 1 year. The secondary data of all confirmed cases of thalassemia were examined by the investigators.

The information regarding sociodemographic details of the patients, type of thalassemia, risk factors such as the family history of genetic disorders, history of consanguinity among parents, symptoms, signs, and complications associated with thalassemia, hematological reports, and management practices was recorded in a predesigned validated proforma.

This retrospective study involved patients who were admitted in the Department of Pediatrics, Jawaharlal Nehru Medical College and Hospital, Bhagalpur, Bihar, India for the purpose of blood transfusion and for getting oral iron chelating agents. Ethical clearance was taken from ethical society.

Parents or guardians were informed about the purpose of the study. For each patient a detailed history was taken from mother or the attendant. After taking brief history preliminary selection was done, and the purpose to the study was explained in detail to its subject. After taking consent from the parents, data was collected, which included sex, age at presentation, age at diagnosis and clinical symptoms at presentation. A thorough physical examination was done in each patient.

Results:

Out of the 80 thalassemia cases, 75 belonged to beta-thalassemia major, 3 of beta-thalassemia intermediate, and 2 of beta-thalassemia minor category.

The mean age of cases was 6.2 years. The age at diagnosis ranged from 0.1 to 11 years. The majority of cases were under-fives 31 (38.75%) and were male. History of consanguineous marriage was positive among 2 cases, 1 involving marriages between second-degree relatives and one involving third-degree relatives [Table 1].

Table-1: Epidemiological profile of thalassemia patients

		N	%
Age group(yrs)	<1	16	20
	1.1-5	31	38.75
	5.5-10	18	22.5
	10.1-15	10	12.5
	15.1-20	4	5
	>20	1	1.25
Gender	Male	48	60
	Female	32	40

Socioeconomic status	Above the poverty line	55	68.75
	Below poverty line	25	31.25
Residence	Urban	69	86.25
	Rural	11	13.75

Fever was the most common presenting symptom 14 (17.5%). Pallor 77 (96.25%) followed by hepatomegaly 75 (93.75%) were the most common signs among cases. Bone deformities were reported in 17 (21.25%) cases, all of which were beta thalassemia major cases [Table 2].

Table-2: Clinical profile of thalassemia patients

Clinical features		N	%
Symptoms	Fever	14	17.5
	Abdominal distension	3	3.75
	Breathlessness	2	2.5
	Diarrhea	2	2.5
	Headache	2	2.5
	Dizziness	4	5
Signs	Pallor	77	96.25
	Hepatomegaly	75	93.75
	Splenomegaly	73	91.25
	Jaundice	12	15
Bone deformities	Bossing of skull	17	21.25
	Hypertrophy of maxilla	9	11.25
	Prominent malar eminence	4	5
	Depression of the nasal bridge	2	2.5

Discussion:

There was a wide spectrum of clinical manifestations among patients of congenital hemolytic anemia. Symptoms of anaemia, pallor, bouts of fever, enlargement of frontal, parietal and maxillary bones (hemolytic facies), and hepatosplenomegaly associated with jaundice and notched ribs are observed for congenital hemolytic anemia. [15-18] Hepatomegaly was the most common clinical finding among the study population (57.8%), followed by splenomegaly (54.9%) and hemolytic facies and jaundice (both 53%). Clinical features of beta thalassemia are usually manifested in younger age group and become more severe with advancing age. HbE beta thalassemia, clinical severity increases with age and complications like those of beta thalassemia eventually develops. Similar results were found in earlier studies. [19-20]

The increased prevalence of HbE Beta Thalassemia in this part of the country was first reported by Chatterjee et al of School of Tropical Medicine, Kolkata in 1966 [21]. Within South Asia, there are about 45 million carriers of beta Thalassemia [22]. Beta thalassaemia among Indian population is seen more commonly in Sindhis, Gujaratis, Bengalis, Punjabis and Muslims[23]. Carrier state for beta

thalassaemia in India varies from 1-17% with a mean of 3.2% [24].

Nearly all children had pallor as a presenting complaint. A small percentage had jaundice. Significant malnutrition was seen in 27 % of patients (Grade 2 and above, as per Indian Academy of Pediatrics classification [25]. Contributing factors to growth retardation include recurrent infections, nutritional deficiency, and chronic hypoxia, iron toxicity from transfusion hemosiderosis, poor transfusional status and inadequate chelation [26-27].

This was per guidelines where the usual frequency of one transfusion every 2–4 weeks is recommended among thalassemia cases so that hemoglobin level is maintained more than 9-10.5 g/dl [28]. This blood transfusion regimen promotes proper growth and prevents bone marrow expansion and iron overload among most patients. The post transfusion hemoglobin also should not be >14–15 g/dl, as it can lead to a greater risk of hyper viscosity and stroke [29]. In this study, the mean post transfusion level of hemoglobin was 10 g/dl indicating appropriate transfusion practices as per guidelines. Furthermore, transfusion in severe thalassemia genotypes as per the guidelines usually starts within the first 2 years of life. Folic acid deficiency has been reported in thalassemia major and intermedia because of increased erythropoiesis. [30]

Conclusion:

Hemoglobinopathies are the commonest hereditary disorders in India and pose a major health problem. The data on the prevalence of β-thalassemia's and other hemoglobinopathies in different caste/ethnic groups of India is scarce.

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