e-ISSN: 0976-822X, p-ISSN:2961-6042

Available online on http://www.ijcpr.com/

International Journal of Current Pharmaceutical Review and Research 2023; 15(10); 96-99

Original Research Article

A Retrospective Record-Based Clinico-Epidemiological Assessment of Thalassemia Patients: An Observational Study

Suprabhat Ranjan[1], Sheela Sinha[2]

[1]Junior Resident, Upgraded Department of Pediatrics, Patna Medical College and Hospital, Patna, Bihar, India

[2]Professor, Upgraded Department of Pediatrics, Patna Medical College and Hospital, Patna, Bihar, India

Received: 06-01-2023 Revised: 21-02-2023 / Accepted: 22-04-2023

Corresponding author: Dr. Suprabhat Ranjan

Conflict of interest: Nil

Abstract

Aim: Thea aim of the present study was to assess the pattern, clinical presentations, complications, and management practices among thalassemia cases

Material & Methods: This was a retrospective record-based cross-sectional study was conducted in the Upgraded Department of Pediatrics. The secondary data of all confirmed cases of thalassemia were examined by the investigators. 100 patients were included in the study.

Results: The mean age of cases was 6.4 years. The age at diagnosis ranged from 0.1 to 11 years. The majority of cases were between 1.1-5 was 36 (36%) and 64 were male. 70% were above poverty line and 85% belonged to urban area. Fever was the most common presenting symptom 17 (17%). Pallor 98 (98%) followed by hepatomegaly 94 (94%) were the most common signs among cases. Bone deformities were reported in 20 (20%) 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

Thalassemia syndromes are caused by inherited mutations that decrease the synthesis of either alpha or beta globin chains of hemoglobin. Imbalance in globin chain synthesis results in anemia, tissue hypoxia, and red cell hemolysis. [1] 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]

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 %. The prevalence of β-thalassemia trait varies between 3-17% because of consanguinity and caste and area endogamy. [4] Thalassemia syndrome is endemic in Mediterranean basin, Middle East, tropical Africa, Indian subcontinent, and Asia. [1] In India, it is the most common single‑gene disorder. [5]

Every year, ten thousand children with β-thalassemia major are born in India, which constitutes 10% of the total number in the world. [6] Thalassemia affects physical growth and delays maturation. Its management also imposes a huge economic burden on the families of the affected. [7] 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 counselling and prenatal diagnosis. [8]

This study attempts to reveal the clinical and hematological profile of patients with different types of thalassemia admitted in Upgraded Department of Pediatrics, Patna Medical College and Hospital, Patna, Bihar, India

Material & Methods

A retrospective record-based cross-sectional study was conducted on pediatric patients aged 0-15 years in Upgraded Department of Pediatrics, Patna Medical College and Hospital, Patna, Bihar, India in between the duration of one year (Feb 2012 to Jan 2013). 100 patients were included in the study. The secondary data of all confirmed cases of thalassemia were examined by the investigators. The data on socio-demographic profile, relevant clinical history and examination and hematological parameters were assessed. Parents or guardians were informed about the purpose of the study.

Methodology

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

Statistical Analysis

Data were entered and analyzed using SPSS Inc., Chicago, IL version 11.0. Chi‑square test, Fisher's exact test, paired t‑test, and Mann–Whitney U‑test were used to test association between variables. Two‑tailed $P < 0.05$ was considered statistically significant association.

Results

Table 1: Epidemiological profile of thalassemia patients

Variables		N	%
Age group(yrs)	<1	20	20
	1.1-5	36	36
	5.5-10	24	24
	10.1-15	14	14
	15.1-20	4	4
	>20	2	2
Gender	Male	64	64
	Female	36	36
Socioeconomic status	Above the poverty line	70	70
	Below poverty line	30	30
Residence	Urban	85	85
	Rural	15	15

The mean age of cases was 6.4 years. The age at diagnosis ranged from 0.1 to 11 years. The majority of cases were between 1.1-5 was 36 (36%) and 64 were male. 70% were above poverty line and 85% belonged to urban area.

Table 2: Clinical profile of thalassemia patients

Clinical features		N	%
Symptoms	Fever	17	17
	Abdominal distension	4	4
	Breathlessness	3	3
	Diarrhea	3	3
	Headache	3	3
	Dizziness	5	5
Signs	Pallor	98	98
	Hepatomegaly	94	94
	Splenomegaly	90	90
	Jaundice	15	15
Bone deformities	Bossing of skull	20	20
	Hypertrophy of maxilla	12	12
	Prominent malar eminence	5	5
	Depression of the nasal bridge	3	3

Fever was the most common presenting symptom 17 (17%). Pallor 98 (98%) followed by hepatomegaly 94 (94%) were the most common signs among cases. Bone deformities were reported in 20 (20%) cases, all of which were beta thalassemia major cases.

Discussion

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. [9] 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. [10-12] 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. [13] 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. [14] Haemoglobinopathies affect 4.5% of the world population. [15]

The mean age of cases was 6.4 years. The age at diagnosis ranged from 0.1 to 11 years. The majority of cases were between 1.1-5 was 36 (36%) and 64 were male. 70% were above poverty line and 85% belonged to urban area. Fever was the most common presenting symptom 17 (17%). Pallor 98 (98%) followed by hepatomegaly 94 (94%) were the most common signs among cases. Bone deformities were reported in 20 (20%) cases, all of which were beta thalassemia major cases. 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. [16,17] 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. [18] Contributing factors to growth retardation include recurrent infections, nutritional deficiency, and chronic hypoxia, iron toxicity from transfusion hemosiderosis, poor transfusional status and inadequate chelation. [19,20]

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. [21] 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. [22] 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. [23]

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.

References

1. Kumar V, Abbasi AK, Aster JC. Red blood cells and bleeding disorders. In: Robbins and Cotran, Pathologic basis of Disease. 9th ed. Gurgaon: Reed Elsevier India Private Limited; 2010. p. 638, 639, 641.
2. Chattopadhyay K, Biswas R, Bhattacherjee S, Bandyopadhyay R. An epidemiological study on the clinico-hematological profile of patients with congenital haemolytic anemia in a tertiary care hospital of Kolkata. Indian J. Prev. Soc. Med. 2011 Oct;43(4):373.
3. Angastiniotis M, Modell B, Englezos P, Boulyjenkov V. Prevention and control of haemoglobinopathies. Bulletin of the World Health Organization. 1995;73(3):375.
4. Trehan A, Sharma N, Das R, Bansal D, Marwaha RK. Clinic investigational and demographic profile of children with thalassemia major. Indian Journal of Hematology and Blood Transfusion. 2010 Mar; 31:121-6.
5. Lokeshwar MR. Late hony. Surg. Cmde. Dr. Shantilal C. Sheth oration presentation during PEDICON 2006, Delhi, January 6th, 2006. Progress in the management of thalassemia. Indian Pediatr 2006; 43:503‑6.
6. Varawalla NY, Old JM, Sarkar R, Venkatesan R, Weatherall DJ. The spectrum of β-thalassaemia mutations on the Indian subcontinent: the basis for prenatal diagnosis. British journal of haematology. 1991 Jun;78 (2):242-7.
7. Thavorncharoensap M, Torcharus K, Nuchprayoon I, Riewpaiboon A, Indaratna K, Ubol BO, et al. Factors affecting health‑related

quality of life in Thai children with thalassemia. BMC Blood Disord 2010; 10:1.
8. La Nasa G, Caocci G, Argiolu F, Giardini C, Locatelli F, Vacca A, Orofino MG, Piras E, Addari MC, Ledda A, Contu L. Unrelated donor stem cell transplantation in adult patients with thalassemia. Bone Marrow Transplantation. 2005 Dec;36(11):971-5.
9. Management of birth defects and hemoglobin disorders. Report of a joint WHO-March of Dimes meeting. Geneva, Switzerland, 17–19 May 2006.
10. Higgs DR, Thein SL, Wood WG. The molecular pathology of thalassemias. In: Weatherall DJ, Clegg B (eds) The thalassemia syndromes, 4th edn. Blackwell Sciences, Oxford, 2001:133–191.
11. Rund D, Rachmilewitz E. Beta- Thalassemia. N Engl J Med 2005:353:1135–1146
12. Gibbon R, Higgs DR, Olivieri NF, Wood WG. The beta thalassemia. In: Weatherall DJ, Clegg JB (eds) The thalassemia syndromes, 4th edn. Blackwell Science, Oxford, 2001: 288–289.
13. Balgir RS. The burden of haemoglobinopathies in India and the challenges ahead. Current Science. 2000;79(11):1536-47.
14. Chattopadhyay K, Biswas R, Bhattacherjee S, Bandyopadhyay R. An epidemiological study on the clinico-hematological profile of patients with congenital hemolytic anemia in a tertiary care hospital of Kolkata. Indian J Prev Soc Med. 2011;43(4):1-6.
15. Angastiniotis M, Modell B, Englezos P, Boulyjenkov V. Prevention and control of hemoglobinopathies. Bull World Health Organ. 1995;73(3):375-86.
16. Erlandson ME, Brilliant R, Smith CH. Comparison of sixty-six patients with thalassemia major and thirteen patients with thalassemia intermedia including evaluations of growth, development and prognosis. Ann Ny Acad Sci. 1964; 7:727-35.
17. Hazell JW, Modell CB. ENT complications in thalassaemia major. J Laryngol Otol. 1976;90 (9):877-81.
18. Proceedings of the workshop on protein calorie malnutrition ecology and management. Indian Pediatr. 1975; 12:57–117.
19. Nutrition subcommittee of the Indian Academy of Pediatrics Report. Indian Pediatr 1972:9:360–372
20. Eshghi P, Alavi S, Ghavami S, Rashidi A (2007) Growth impairment in beta thalassemia major: the role of trace element deficiency and other potential factors. J Pediatr Hematol Oncol 29(1): 5–8
21. Espinosa M. F. M., Erazo E. W. V., Villada N. Z., Sánchez D. A. G., García, J. S. R., Peña C. A. E., Mejía A. O., Rey, J. V., & Pertuz, J. G. V. Treatment of Ventral Hernia Laparoscopic or Open Approach? Journal of Medical Research and Health Sciences, 2022:5(4), 1876– 1880.
22. Louis CK. Growth of children with beta thalassemia major. Indian J Pediatr 2005:72(2) :159–164
23. Mallik S, Chatterjee C, Mandal PK, Sardar JC, Ghosh P, Manna N. Expenditure to treat thalassaemia: an experience at a tertiary care hospital in India. Iran J Public Health. 2010;39(1):78-84. Epub 2010 Mar 31.