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Clinical Profile of Children with Thalassemia Admitted for Blood Transfusion at a Tertiary Health Care Center

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ABSTRACT

Introduction

Thalassemia is a prevalent hereditary hematological disorder that is characterized by a reduction or absence of globin chain synthesis. The objective of this study is to determine the clinical profile of children with thalassemia who undergo repeated packed cell transfusions.

Methods

A prospective cross-sectional observational study was conducted from January 1, 2023, to May 31, 2023, in the Pediatric ward at Bharatpur Hospital. Ethical clearance was obtained from the Institutional Review Committee of Bharatpur Hospital (Ref: 078/79-018). A total of 53 cases of Thalassemia, who were receiving regular blood transfusions, were included in this study.

Results

The prevalence of thalassemia was 3.68% (with 95% CI 2.7% to 4.65%). The average age at diagnosis was 15.58 months, with a range of 2 to 97 months. The majority of cases, 32 (60.37%), were diagnosed before the age of one year, with a male-to-female ratio of 1.2:1. Thalassemia was more prevalent in the Tharu community (58.5%) compared to other communities.

Conclusions

Thalassemia is a hematological disorder that predominantly affects the Tharu community. Common clinical manifestations of transfusion-dependent thalassemia in children include pallor, hepatomegaly, splenomegaly, and facial deformities. Therefore, it is imperative to monitor serum ferritin levels to detect iron overload early and initiate chelation therapy promptly to prevent complications. Such measures can significantly improve the quality of life of affected individuals and reduce the burden of this genetic disorder on affected communities.

Keywords: blood transfusion; chelation therapy; ferritin levels; hematological disorders; thalassemia.

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INTRODUCTION

Thalassemia is a prevalent hereditary hematological disorder characterized by a reduction or absence of globin chain synthesis.[1,2] A diverse range of genetic defects underlies this condition.[3] The global incidence of thalassemia carriers is estimated to be approximately 7% of the world's population.[4] Notably, the Tharu community in Nepal has a higher prevalence of thalassemia gene carriers.[5] Beta thalassemia in young children typically manifests within the first year of life[4]. Severe thalassemia is associated with distinct facial features, marked hepatosplenomegaly, and cachexia.[6, 7] The objective of this study is to establish the profile of thalassemia children who require repeated packed cell transfusions, with the aim of initiating early chelation therapy to minimize the morbidity and mortality associated with iron overload. Therefore, thalassemia patients receiving blood transfusions at Bharatpur Hospital were assessed and advised on appropriate chelation therapy.

METHODS

A cross-sectional study was conducted in the Pediatric ward of Bharatpur Hospital, Nepal, from January 1, 2023, to May 31, 2023. Ethical clearance was taken from Institutional review committee of Bharatpur Hospital (Ref: 078/79-018). Informed and written consent was taken from parents. The study included known cases of thalassemia who were receiving regular blood transfusions in the Pediatrics department at Bharatpur Hospital. Children of age above 14 years and who were receiving irregular transfusions were excluded from the study. Self-structured questionnaire were used to collect information on the children's sex, age at diagnosis, current age, and time interval for transfusion. Additionally, the children's caste was recorded from both departmental records and parental reports. Physical examinations, including liver span, spleen span, and facial deformities, were conducted and recorded. Data were entered and analyzed using IBM SPSS Statistics version 18. Data was analyzed by using descriptive statistical tools. In the descriptive statistics mean and SD was calculated for continuous variable while frequency and percentage were calculated for categorical variables.

RESULTS

From January 1, 2023, to May 31, 2023, 1438 cases were admitted in pediatric ward of Bharatpur Hospital among them 53 cases of Thalassemia, who were receiving regular blood transfusions. This showed that the prevalence of thalassemia was 3.68% (with 95% CI 2.7% to 4.65%) (Table 1).

Table 1. Prevalence and 95% CI of Thalassemia.

Thalassemia	Number of patients	Percentage	95% CI	
			Lower	Upper
Yes	53	3.68	2.7	4.65
No	1385	96.32		

The mean age of the children was 15.58 months, with a range spanning from 2 to 97 months. Among the 53 cases examined, 29 (54.7%) were male and 24 (45.3%) were female. The prevalence of thalassemia was found to be higher in the Tharu community in comparison to other communities. This was evidenced by the fact that out of the 53 cases observed, 31 (58.5%) were from the Tharu community (Table 2).

Table 2. Demographic characteristics of children with thalassemia (n=53).

Variables	Number (%)
Age	
<5	12 (22.64)
5-9	18 (33.96)
10-14	23 (43.4)
Gender	
Male	29 (54.7)
Female	24 (45.3)
Caste	
Tharu/Chaudhary	31(58.5)
Magar	8(15.0)
Brahamin	4(7.5)
Darai	5(9.5)
Kumal	3(5.7)
Others	2 (3.8)

The majority of cases, 32 (60.37%), were diagnosed before the age of one year. The primary presenting symptom among children was progressive pallor, while other notable findings included hepatomegaly, splenomegaly, icterus and facial dysmorphism. The majority of cases 27 (50.9%) received blood transfusions biweekly, while 10 (18.9%) received transfusions every three weeks, and 15 (28.3%) received transfusions every four weeks. Only one case received transfusions every five weeks. Of the 53 cases, 7(13.2%) had serum ferritin levels below 1000 ng/ml, while 37 (69.8%) had levels between 1000 and 2500 ng/ml, and 9 (17%) had levels above 2500 ng/ml. This research showed that 21 (39.63%) were undergoing regular iron chelation therapy while, 2 (3.77%), were receiving irregular chelation therapy. The remaining 30 (56.6%) of the children, were not undergoing any form of chelation therapy (Table 3).

Table 3. Clinical profile of patients (n=53).

Variables	Number (%)
Age at diagnosis (months)	n (%)
0 - 6	17 (32.1)
7 - 12	15 (28.3)
13 - 18	6 (11.3)
19 - 24	9 (17.0)
≥25	6 (11.3)
Signs	
Pallor	53 (100)
Icterus	16 (30.2)
Hepatomegaly	48 (90.5)
Spleenomegaly	47(88.7)
Facial Dysmorphism	34 (64.2)
Interval between transfusion (Weekly)	
2	27 (50.9)
3	10 (18.9)
4	15 (28.3)
5	1 (1.9)
Serum ferritin level	
< 1000	7 (13.2)
1001 - 2500	37 (69.8)
≥2500	9 (17)
Chelation therapy	
No	30 (56.6)
Regular	21 (39.63)
Irregular	2 (3.77)

DISCUSSIONS

In the current investigation, a male predominance was observed, with a male-to-female ratio of 1.2:1. This finding is consistent with a study conducted in India in 2016, which reported a male predominance in thalassemia prevalence, with 53.3% of males and 46.6% of female affected.[8] Similarly, a study from Tunisia found a higher percentage of thalassemia cases in males (55.4%). [9] In the present study, the

majority of children were diagnosed before infancy, which is in line with the findings of Modella and Berdukas.[2] The mean age of the children in the present study was 8.53 ±3.96 years, with 43.4% of children falling within the 10 to 14 years age group. This result is consistent with a study conducted in Pakistan by Riaz et al., where the mean age was 10.8 ± 4.5 years and 46.5% of children were between the ages of 10 and 14 years.[11] The mean age at diagnosis in the present study was 15.58 months, with a range of 2 to 97 months.

In the present study, 31 cases (58.5%) were identified as belonging to the Tharu population. While thalassemia is more commonly observed in the Tharu community, cases were also found in the Magar, Darai, and Brahamin populations. This highlights the importance of mandatory screening and genetic counseling for all individuals, regardless of their caste. A study conducted by H C Upreti in 2020 similarly found a higher prevalence of thalassemia in Tharu communities.[5] All cases in the present study presented with progressive pallor, which prompted their need for blood transfusion. A study conducted by A Trehan similarly found that progressive pallor was observed in the majority of cases (99%).[4] In the present study, the presenting features for blood transfusion were facial dimorphisms, Icterus, Hepatomegaly, and Splenomegaly. These findings are consistent with those reported in a study conducted by A Trehan.[4]The majority of children with thalassemia, 27 (50.9%), required regular blood transfusions every two weeks. However, this regular hospital visit imposes a financial burden on families and increases the risk of transfusion-related complications in children. The measurement of ferritin levels in the blood is a reliable method for assessing iron overload in the body, which is generally indicative of total iron levels. Among the children studied, 37 (69.8%) exhibited serum ferritin levels ranging from 1001 to 2500 ng/ml. Of the remaining cases, 9 (17%) exhibited levels exceeding 2500 ng/ml, while only 7 (13.2%) exhibited levels below 1000 ng/ml. Notably, those who underwent regular chelation therapy exhibited lower ferritin levels. These findings are consistent with those of a study conducted by AK Mishra, which similarly found that the majority of cases exhibited ferritin levels exceeding 1000 ng/ml, with only 12.5% exhibiting levels below 1000 ng/ml.[10] In the current investigation, a mere 21 (39.63%) children were receiving consistent chelation therapy, and this was sourced from an external facility. The lack of access and financial constraints associated with chelation therapy were identified as significant factors contributing to iron overload and transfusion-related complications in thalassemic children. This observation is consistent with the findings of a previous study conducted by Sharma and Poudyal B.[12]

CONCLUSIONS

Thalassemia is a hereditary condition that is prevalent among the Tharu community in Nepal. Children with transfusion-dependent thalassemia often exhibit symptoms such as pallor, hepatomegaly, splenomegaly, and facial deformities. Therefore, it is crucial to regularly monitor serum ferritin levels to detect iron overload early and initiate chelation therapy promptly to prevent complications. The provision of community education, genetic counseling, and access to transfusion and early chelation therapy complication , timely detection, management and genetic counselling can significantly improve the quality of life for affected children.

Limitations

Limitations of this study include the fact that only pediatric patients who presented for transfusion were included, which may not fully reflect the burden of cases within the broader community.

REFERENCES

1. Jha R, Jha S. Beta thalassemia-a review. Journal of Pathology of Nepal. 2014 Sep 24;4(8):663-71.
2. Shafique F, Ali S, Almansouri T, Van Eeden F, Shafi N, Khalid M, Khawaja S, Andleeb S. Thalassemia, a human blood disorder. Brazilian Journal of Biology. 2021 Sep 3;83.
3. Scianò F, Bramanti B, Gualdi-Russo E. A new investigative strategy to diagnose β-thalassemia syndrome in past human populations. Archaeological and Anthropological Sciences. 2021 Feb;13:1-2.
4. Trehan A, Sharma N, Das R, Bansal D, Marwaha RK. Clinicoinvestigational and demographic profile of children with thalassemia major. Indian Journal of Hematology and Blood Transfusion. 2015 Mar;31:121-6.
5. Shrestha RM, Pandit R, Yadav UK, Das R, Yadav BK, Upreti HC. Distribution of hemoglobinopathy in Nepalese population. J Nepal Health Res Counc. 2020 Apr 20;18(1):52-8.
6. Ali MR, Bari MI, Mia MS, Rahman MK, Hossain MF, Sharmin LS. Clinical Profile of Thalassemia Syndrome in Children of Northern Bangladesh. TAJ: Journal of Teachers Association. 2018;31(2):6-11.
7. Shah N, Mishra A, Chauhan D, Vora C, Shah NR. Study on effectiveness of transfusion program in thalassemia major patients receiving multiple blood transfusions at a transfusion center in Western India. Asian Journal of transfusion science. 2010 Jul;4(2):94.
8. Choudhari S, Behera MK. Chelation status & clinical profile of thalassemic children attending paediatric clinics. Indian Journal of Applied Researc. 2016;6:276-8.
9. BejaouiM,GuiratN.BETATHALASSEMIA MAJOR IN A DEVELOPING COUNTRY: EPIDEMIOLOGICAL, CLINICAL AND EVOLUTIONARY ASPECT. Mediterranean Journal of Hematology and infectious diseases. 2013 Jan 1;5(1):e2013002-.
10. Mishra AK, Tiwari A. Iron overload in beta-thalassemia major and intermedia patients. Maedica. 2013 Sep;8(4):328.
11. Riaz H, Riaz T, Khan MU, Aziz S, Ullah F, Rehman A, Zafar Q, Kazi AN. Serum ferritin levels, socio-demographic factors and desferrioxamine therapy in multi-transfused thalassemia major patients at a government tertiary care hospital of Karachi, Pakistan. BMC Res Notes. 2011 Aug 11;4:287. doi 10.1186/1756-0500-4-287. PMID: 21834969; PMCID: PMC3162919
12. Sharma Poudyal B, Devkota A, Kouides P. Thalassemia care in Nepal: In dire need of improvement. eJHaem.

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