Hemoglobinopathies in Children with Anemia Presenting to Nepalgunj Medical College

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ABSTRACT

Background

Anemia refers to reduced number of red blood cells or haemoglobin level lower than the standard range for age and sex. It is a major global health issue and is an indicator of poor nutrition, health as well as the social and economic development of a population. Symptoms are lethargy, fatigue, weakness, and shortness of breath. Causes include iron deficiency, vitamin deficiencies, malaria, intestinal helminthes, viral infections, chronic diseases, hemoglobinopathies, hemolysis etc. Hemoglobinopathies are the most prevalent single-gene autosomal recessive disorders which involve disorders in globin chain synthesis, that may either be quantitative (thalassaemias) or qualitative (sickle cell). These disorders can be prevented by proper genetic counselling and screening.

Objective

To find the prevalence and distribution of hemoglobinopathies among anemic children attending Nepalgunj Medical College.

Method

The cross-sectional study was carried out in the department of Pediatrics, on 86 children, aged 12 months - 15 years, in whom anemia was detected, either on examination or on investigation. Complete blood counts, peripheral blood smear, and Hb-electrophoresis were done.

Result

Among 86 children, majority were of age group 1 - 5 years (40.7%), making male : female ratio 1.2:1. The youngest child was of 12 months with average age 8.0 ± 4.8. Commonest morphology was microcytic hypochromic anemia (81.4%). Hemoglobinopathies was seen in 31.39% children, sickle cell disease being most common (16.3%), followed by Beta Thalassemia trait (4.7%), 73.3% children were from tharu family. There was no statistically significant association between ethnicity and hemoglobinopathies as well as sickle cell disease.

Conclusion

Hemoglobin disorders are also common causes of anemia in children. These were seen in various ethnic groups and were not limited to tharus with consanguineous marriages. Thus, hemoglobinopathies should also be thought-of in non-tharu people with microcytic hypochromic anemia. However, further studies on larger population are needed to determine significant relation between ethnicity and hemoglobinopathies.

KEY WORDS

Anemia, Hemoglobinopathies, Sickle cell anemia, Thalassemia, Tharu

INTRODUCTION

Anemia is defined as reduced number of red blood cells or hemoglobin level lower than the standard range for age and sex. It is a major global health issue and leading contributor to illness and deaths among children.[1] It reflects poor nutrition, poor health status, and the social and economic development of a population.[2] Common symptoms are lethargy, fatigue, weakness, and shortness of breath. According to the World Health Organization, the global prevalence of anemia in children aged 6–59 months is approximately 39.8%.[3] Causes are iron deficiency, folate, vitamin A and B12 deficiencies, malaria, intestinal helminthes, viral infections, chronic diseases, hemoglobinopathies, hemolysis, and bone marrow disorders.[4] Majority of them, show microcytic hypochromic anemia, common causes being iron deficiency and thalassemia.[5-7]

Hemoglobinopathies are the most prevalent single-gene autosomal recessive disorders globally, with wide geographical variation. They are the disorders of globin chain synthesis, that may either be quantitative (thalassaemias) or qualitative (Sickle cell, Hb C, Hb E).[8] In Southeast Asia, α-thalassemia, β-thalassemia, Hb E, and Hb S are common.[9]

Thalassemia is characterized by reduced or absent production of one or more globin chains forming hemoglobin tetramer. Sickle cell disease, the first human disease whose mechanism was understood at the molecular level, results due to missense mutation leading to substitution of glutamate with valine amino acid in the globin chain of hemoglobin.[10]

Thalassemia and sickle cell anemia can be prevented by proper genetic counselling and screening.[11] Thus, this study aims to assess the prevalence and distribution of hemoglobinopathies among anemic children attending Nepalgunj Medical College.

METHODS

The cross-sectional study was carried out in the department of Pediatrics at Nepalgunj Medical College Teaching Hospital, Kohalpur from July 2025 to February 2026. Ethical clearance was obtained from the Institutional Review Committee, Nepalgunj Medical College and Teaching Hospital. The study included 86 children, aged 12 months to 15 years, attending Pediatric Department in whom anemia was detected, either on examination or on investigation (according to WHO guidelines). Selection of cases was done by the convenient sampling method. After obtaining written informed consent, detailed relevant history, and clinical examination, a blood sample by vein puncture was collected for complete blood counts (if not done earlier), peripheral blood smear, and Hb-electrophoresis.

Those younger than 12 months or older than 15 years, previously treated for anaemia, previously diagnosed with haemoglobinopathy, or suffering from chronic diseases were excluded.

Data were collected in a predesigned proforma, which included demographic informations such as age, sex, address and caste, haemoglobin level, type of anaemia and haemoglobinopathies diagnosed. The results were tabulated and statistically analyzed using SPSS version 25.

RESULTS

During the study period, anemia was detected in 86 children. Among them, 47 (54.7%) were males and 39 (45.3%) were females, giving a male-to-female ratio of 1.2:1. Majority of the children were of age group 1-5 years (40.7%), followed by children of 10-15 years (39.5%). Rest 19.8% were of age group 6-10 years. The youngest child was of 12 months and average age was 8.0 ± 4.8 years.

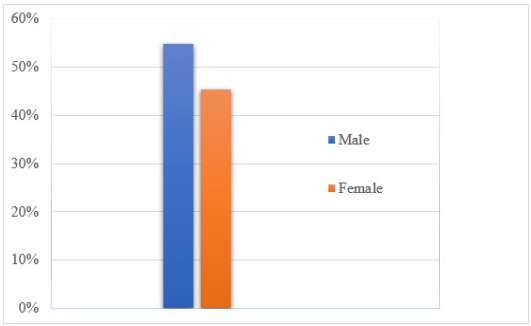

Figure 1. Sex distribution of the study population

Consanguineous marriage in the parents of children with anemia under study was present in 9.3% while in majority (90.68%) it was non-consanguineous.

On the basis of morphology, microcytic hypochromic anemia was found in 70 (81.4%) children, macrocytic anemia was in 5 (5.8%) cases while rest 11 (12.8%) had normocytic normochromic anemia, as shown in the table 1.

Table 1. Morphological variation of anemia

Morphology	Number (N)	Percentage (%)
Microcytic hypochromic	70	81.4
Normocytic normocytic	11	12.8
Macrocytic hypochromic	5	5.8
Total	86	100

Hemoglobinopathies was seen in 27 (31.39%) children during the study period. Most common hemoglobinopathy was sickle cell disease, 14 (16.3%), followed by Beta Thalassemia trait (4.7%), as shown in the table 2.

Table 2. Hemoglobinopathies in the anemic children

Hemoglobinopathy	Number (N)	Percentage (%)
Sickle cell disease	14	16.3
Sickle cell anemia	8	9.3
Sickle cell trait	6	6.9
β-thalassemia trait	4	4.7
Others	9	10.4
Compound heterozygous for sickle cell and β-thalassemia	7	8.1
Hereditary persistence of HbF	2	2.3

Among the study population, 63 (73.3%) children belonged to Tharu family while 23 (26.7%) were non-tharu. There was no statistically significant association between ethnicity and hemoglobinopathies (Table 3) as well as sickle cell disease (including sickle cell disease and compound heterozygous for sickle cell and β-thalassemia) (Table 4).

Table 3. Relation of ethnicity of the study population with hemoglobinopathies

Tharu	Hemoglobinopathies			p-value
	No	Yes	Total	
No	17	6	23	0.35
Yes	42	21	63	
Total	59	27	86	

Table 4. Relation of ethnicity of the study population with sickle cell disease

Tharu	Sickle Cell Disease			p-value
	No	Yes	Total	
No	19	4	23	0.26
Yes	46	17	63	
Total	65	21	86	

DISCUSSIONS

Anaemia in children is a global public health issue, particularly in low- and middle-income countries. It has been associated with visual and auditory dysfunctioning, cognitive, behavioral abnormalities, and delay in psychomotor development.[12] Therefore, appropriate screening and diagnosis at the earliest can prevent significant morbidity and mortality.

The sex distribution in the present study showed male predominance 54.7% with male-to-female ratio of 1.2:1, consistent with the study by Jui et al. and Sanghvi et al.[11,13] This might be the result of socio-cultural beliefs in certain communities where male children receive more attention and healthcare access. Anemia was more prevalent in younger children, 40.7% in 1-5 years, followed by 10-15 years (39.5%) and 19.8% in children of 6-10 years. This is similar to the study by Colah et al. and Sanghavi et al.[11,14] This shows younger children are vulnerable to dietary nutritional deficiencies and have higher nutritional demands for rapid growth.

Consanguineous marriage in the parents of children with anemia under study was present in 9.3% while in majority (90.68%) it was non-consanguineous. In concordance to the studies by Sameer et al. and Choudhary et al. where majority of marriages were non-consanguineous.[15,16]

Morphologically, in the present study, microcytic hypochromic anemia was most common (81.4%), followed by normocytic normochromic anemia (12.8%) and macrocytic anemia was present in 5.8% children. These findings were similar to the studies by, Saba et al. and Kapoor et al. in which microcytic hypochromic anemia was most common followed by normocytic normochromic and the least common was macrocytic anemia.[12,17]

In the present study, prevalence of hemoglobinopathy was 31.39% children. The study by Mondal et al. had 35.0% whereas the study by Warghade et al. for abnormal Hb showed rate of 18.44% as they included large population of all ages from all over India to identify abnormal Hb whereas our study included a small number of children, in whom anemia was detected during examination or on investigation.[18,19]

The most common hemoglobinopathy detected, in the study, was sickle cell disease (16.3%), followed by Beta Thalassemia trait (4.7%), and very few children had other rare hemoglobinopathies like compound heterozygous for sickle cell and β-thalassemia (8.1%), hereditary persistence of HbF (2.3%). In the studies by Mondal et al. and Sanghavi et al. also most prevalent hemoglobinopathy was sickle cell disease.[11,18] However, Warghade et al. in their study, found beta thalassemia trait as the most prevalent hemoglobinopathy.[19] This variation in hemoglobinopathies might be because of different study population and places. Cluster of genes causing these defects might vary by place and population.

Among the study population, in the study, 73.3% children were of tharu ethnicity while 26.7% were from non-tharu family. There was no statistically significant relation between hemoglobinopathies and tharu race. Also, no statistical significance was found between sickle cell disease (including sickle cell disease and compound heterozygous for sickle cell and beta thalassemia) and tharu ethnicity.

Different studies done in various parts of Nepal, India and Pakistan have shown sickle cell disease and other hemoglobinopathies, more prevalent in tharu people but none of the studies have shown statistically significant association with them.[20-25] Further studies with large number of study population are warranted to establish statistically significant association between tharu ethnicity and hemoglobinopathies. Relatively small sample size and convenient sampling method in the study might have led to selection bias. Hb-electrophoresis was used for diagnosis, molecular/genetic testing was not performed, which

might have limited the precise diagnosis and potentially altered the results. The study was single-center-hospital based study. Therefore, the results cannot be generalized to children in other regions of Nepal. Larger, multicentric studies are recommended to confirm these findings.

CONCLUSION

This study shows that hemoglobin disorders are also common causes of anemia in children, prevalent in about 31.39%. This highlights the importance of early detection and intervention. Sickle cell disease and β-thalassemia trait were the most common, especially in younger children. These conditions were seen in various ethnic groups and were not limited to certain groups (tharu) with consanguineous marriages. Thus, hemoglobinopathies should also be thought of in non-tharu population who have microcytic hypochromic anemia and normal iron profile. However, further studies involving larger population are needed to determine statistically significant association between tharu ethnicity and hemoglobinopathies.

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