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Assessment of Clinico- Haematological Profile of Hb E-Beta Thalassemic Children

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Abstract
Background: The aim is to evaluate clinico- haematological profile of Hb E-Beta Thalassemic children. **Material and Methods:** Fifty- six Hb E-Beta Thalassemic children were enrolled in present study. 5 ml of venous blood was collected and analysed in fully automated counter. RBC, WBC, MCV, MCH, MCHC, platelet count, reticulocyte count was assessed. Staining of peripheral smear was done using Leishman stain. High performance liquid chromatography was used for assessment of haemoglobin type. **Results:** Out of 56 children, boys were 30 and girls were 26. The mean spleen size was 4.3 cm in mild, 4.6 cm in moderate and 6.2 cm in severe form. The mean Hb was 7.1 gm/dl, 6.5 gm/dl and 6.2 gm/dl. The mean MCV was 68.6 fl, 68.2 fl and 64.9 fl. The mean MCH was 19.3 pgm, 19.0 pgm and 19.6 pgm. The mean RDW was 31.5 CV%, 31.2 CV% and 32.8 CV%. The mean HB F was 37.2%, 33.5% and 32.8%. The mean HB E was 59.4%, 48.7% and 59.1%. Clinical scoring was 2.7 in mild, 5.2 in moderate and 8.3 in severe form. Clinical symptoms were pallor in 50, jaundice in 25, hepatomegaly in 22, splenomegaly in 42 and hemolytic facies in 35 patients. The difference was non- significant (P>0.05). **Conclusion:** Patients with Hb E thalassemia have variable clinical and hematological profile.
Keywords: β thalassemia, Children, Hemoglobin E.

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Introduction

Hemoglobin E (HbE) is variant hemoglobin with a mutation in the globin gene causing substitution of glutamic acid for lysine at position 26 of the globin chain. HbE is the second most common structural hemoglobin disorder after sickle cell hemoglobin (HbS).[1] It is common in South-East Asia, with a prevalence of as high as 30-40% in some parts of Thailand and Cambodia and in Laos. Hb E is also commonly found in Sri Lanka, North Eastern India, Bangladesh, Pakistan, Nepal, Vietnam, and Malaysia.[2] HbE may be present in three different forms: Heterozygous state (genotype AE or hemoglobin E trait), Homozygous state (EE or hemoglobin E disease) or Compound heterozygous states such as hemoglobin E/β Thalassemia (E/β thal), sickle cell/hemoglobin E disease (SE genotype).[3]

The prevalence of β- thalassemia trait varies between 3- 17% because of consanguinity and caste and area endogamy. Every year, ten thousand children with β thalassemia major are born in India, which constitutes 10% of the total number in the world. HbE thalassemia is common in north-east parts of India.[4] The only forms of treatment available for thalassemia patients are regular blood transfusion, iron chelation therapy in an attempt to prevent iron

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overload and the judicious use of splenectomy in cases complicated by hypersplenism.[5] 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.[6] We performed present study with the aim to evaluate clinico- hematological profile of Hb E-Beta Thalassemic children.

Material and Methods

Fifty- six Hb E-Beta Thalassemic children were enrolled in present study after obtaining approval from ethical review committee of the institute.

Parameters such as onset, duration, and frequency of presenting symptoms were recorded. Icterus, pallor, haemolytic facies, skin changes, hepato- splenomegaly were recorded. 5 ml of venous blood was collected and analysed in fully automated counter. RBC, WBC, MCV, MCH, MCHC, platelet count, reticulocyte count was assessed. Staining of peripheral smear was done using Leishman stain. High performance liquid chromatography was used for assessment of haemoglobin type. PCR was used for detecting the polymorphisms of selected gene. The beta globin gene mutations were identified by ARMS- PCR. The results were compiled and subjected for statistical analysis using Mann Whitney U test. P value less than 0.05 was set significant.

Results

Table 1: Patients distribution

Total- 56		
Gender	Boys	Girls
Number	30	26

Out of 56 children, boys were 30 and girls were 26 [Table 1].

Table 2: Assessment of clinico- hematological profile

Parameters	**Mild**	**Moderate**	**Severe**	**P value**
Spleen size (cm)	4.3	4.6	6.2	0.41
Hb (gm/dl)	7.1	6.5	6.2	0.34
MCV (fl)	68.6	68.2	64.9	0.95
MCH (pgm)	19.3	19.0	19.6	0.12
RDW (CV%)	31.5	31.2	32.8	0.31
HB F (%)	37.2	33.5	32.8	0.92
HB E (%)	59.4	48.7	59.1	0.90
Clinical scoring	2.7	5.2	8.3	0.81

The mean spleen size was 4.3cm in mild, 4.6 cm in moderate and 6.2 cm in severe form. The mean Hb was 7.1 gm/dl, 6.5 gm/dl and 6.2 gm/dl. The mean MCV was 68.6 fl, 68.2 fl and 64.9 fl. The mean MCH was 19.3 pgm, 19.0 pgm and 19.6 pgm. The mean RDW was 31.5 CV%, 31.2 CV% and 32.8 CV%. The mean HB F was 37.2%, 33.5% and 32.8%. The mean HB E was 59.4%, 48.7% and 59.1%. Clinical scoring was 2.7 in mild, 5.2 in moderate and 8.3 in severe form. The difference was non- significant (P>0.05) [Table 2].

Table 3: Clinical parameters

Symptoms	**Number**	**P value**
Pallor	50	0.12
Jaundice	25	

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Hepatomegaly	22	
Splenomegaly	42	
Hemolytic facies	35	

Clinical symptoms were pallor in 50, jaundice in 25, hepatomegaly in 22, splenomegaly in 42 and hemolytic facies in 35 patients. The difference was non- significant (P>0.05) [Table 3].

Discussion

The World Health Organization (WHO) recognized thalassemia as the most prevalent genetic blood disorder in the world, found in more than 60 countries with a carrier population of up to 150 million.[7] This disorder is highly prevalent among children in the Middle East, Mediterranean region, and South Asia.[8] Thalassemia is a chronic disease and presents with a wide range of serious clinical and psychological challenges.[9,10] The effects of thalassemia on physical health can lead to physical deformity, growth retardation, and delayed puberty.[11] Its impact on physical appearance, e.g., bone deformities and short stature, also contributes to a poor self-image.[12] We performed present study with the aim to evaluate clinico-hematological profile of Hb E-Beta Thalassemic children.

We found that out of 56 children, boys were 30 and girls were 26. Kumar et al,[13] found that out of 211 patients evaluated, most common cause of congenital hemolytic anemia was Hb E Beta thalassemia (39.8%), followed by beta thalassemia (27.9%), beta thalassemia trait (14.2%), Hb E disease (11.3 %) and Hb E trait (6.6%). There was male preponderance (male 63%, female 37 %). The mean hemoglobin was found to be lowest in patients of β thalassemia (5.1 gm/gl) and HbE β thalassemia (5.8 gm/dl). The mean total serum bilirubin was found to be highest among β Thalassemia patients (3.0 mg/dl). 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%). The incidence of HbE beta thalassemia is relatively high in comparison to other varieties of thalassemias and is a major public health problem in this area of the country

We found that the mean spleen size was 4.3 cm in mild, 4.6 cm in moderate and 6.2 cm in severe form. The mean Hb was 7.1 gm/dl, 6.5 gm/dl and 6.2 gm/dl. The mean MCV was 68.6 fl, 68.2 fl and 64.9 fl. The mean MCH was 19.3 pgm, 19.0 pgm and 19.6 pgm. The mean RDW was 31.5 CV%, 31.2 CV% and 32.8 CV%. The mean HB F was 37.2%, 33.5% and 32.8%. The mean HB E was 59.4%, 48.7% and 59.1%. Clinical scoring was 2.7 in mild, 5.2 in moderate and 8.3 in severe form. We found that clinical symptoms were pallor in 50, jaundice in 25, hepatomegaly in 22, splenomegaly in 42 and hemolytic facies in 35 patients. Ismail et al,[14] evaluated the health-related quality of life (HRQoL), muscular strength and pain in children with β-thalassemia major. One hundred and twenty children (60 with β-thalassemia major and 60 age-matched healthy) were participated in a cross-sectional study from both sexes (57 girls and 63 boys) with ages ranging from two to twelve years. HRQoL (physical, emotional, social and school functioning), muscular strength and pain were evaluated for all children by using the pediatric quality of life inventory™ (PedsQL™) 4.0 generic core scale, hand-held dynamometer and visual analogue scale (VAS) respectively. Children with β-thalassemia major showed a significant decrease in all domains of health-related quality of life and handgrip strength with a significant increase in VAS score.

Conclusion

Patients with Hb E thalassemia have variable clinical and hematological profile.

References

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1. Angastiniotis M, Modell B, Englezos P, Boulyjenkov V. Prevention and control of hemoglobinopathies. Bull World Health Organ. 1995;73(3):375-86.
2. Balgir RS. The genetic burden of hemoglobinopathies with special reference to community health in India and the challenges ahead. Indian J Hematology Blood Transfusion. 2002;20(1):2-7.
3. Varawalla NY, Old JM, Sarkar R, Venkatesan R, Weatherall DJ. The spectrum of beta thalassemia mutations on the Indian subcontinent: the basis for prenatal diagnosis. Brit J Hematol. 1991;78(2):242- 7.
4. Mohanty D, Colah RB, Gorakshakar AC, Patel RZ, Master DC, Mahanta J, et al. Prevalence of β-thalassemia and other haemoglobinopathies in six cities in India: a multicentre study. J Community Genet. 2013;4(1):33-42. doi: 10.1007/s12687-012-0114-0.
5. Nasa LG, Caocci G, Argiolu F. Unrelated donor stem cell transplantation in adult patients with thalassemia. Bone Marrow Transplant. 2005;36(11):971-5.
6. Chatterjea JB. Some aspects of hemoglobin E and its genetic interference with Thalassemia. Ind J Med Res. 1965;53:377.
7. Sujatha R, Sreekantha, Niveditha SR, Avinash SS, Remya, Vinodchandran, et al. The study of recent biochemical and pathological aspects of thalassemia. Int J Research Health Sci. 2013;1(3):140-52.
8. Mondal B, Maiti S, Biswas BK, Ghosh D, Paul S. Prevalence of hemoglobinopathy, ABO and rhesus blood groups in rural areas of West Bengal, India. J Res Med Sci. 2012;17(8):772-6.
9. Archana AD, Kavita D, Pragna R. Biochemical patterns of hemoglobinopathies and thalassemia syndrome in a tertiary care hospital of Telangana. International J Healthcare Sci. 2014;2(2):385-8.
10. Weatherall DJ, Clegg JB. Thalassemia - a global health problem. Nat Med. 1996;2:847-9.
11. Strauss BS. Genetic counseling for thalassemia in the Islamic Republic of Iran. Perspect Biol Med. 2009;52(3):364-76. doi: 10.1353/pbm.0.0093.
12. 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.
13. Kumar S, Singh D, Garg A. An epidemiological study on the clinico-haematological profile of pediatric patients with congenital hemolytic anemia. Int J Contemp Pediatr. 2017;4:374-7.
14. Caocci G, Efficace F, Ciotti F, Roncarolo MG, Vacca A, Piras E, et al. Health related quality of life in Middle Eastern children with beta-thalassemia. BMC Blood Disord. 2012;12:6. doi: 10.1186/1471-2326-12-6