Thalassaemias and Hemoglobinopathies in a Tertiary Care Hospital, Rangpur

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Thalassaemias are a group of genetic, inherited disorders of the blood that result from a reduced rate of synthesis of α or β chain. More specifically, it is a disorder of hemoglobin molecule inside the red blood cells. According to World health organization there are about 3% beta thalassaemia carrier and about 4% Hb E carrier in Bangladesh. Every year about 300,000 infants worldwide born with thalassaemia syndrome (30%) and sickle cell anaemia (70%). Haemoglobinopathies may be defined as a reduced rate of synthesis of haemoglobin due to structural defect of haemoglobin. Hemoglobinopathies are a major health problem all over the world including India and South Asian region. Identification of these disorders is important for epidemiologic purposes and for prevention of thalassaemia major and clinically severe hemoglobinopathies. Aims of our studies were to see the pattern of hemoglobinopathies and thalassaemias, clinical profiles of the patients, give genetic counseling to the patients. Total 500 patients were selected in our study. Most of the patients (45.2%) were >20 years age. 24.2% were < 10 years of age. Female (51.2%)were more than male (48.8%). Majority of the patients were Muslim (84%). Pattern of the patients were Hb E/beta thalassaemia 40.8%, Hb E trait 30%, Hb E disease 24.8%, Beta thalassaemia trait 3.4%, Beta thalassaemia major 1.2%. Majority patients were anaemic (95%), icteric (60%), splenomegaly (43%), hepatomegaly (19%).

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Introduction

Thalassaemia is the most common genetic blood disease in the world and varies in different population groups in the world.[1] World health organization estimates that at least 6.5% of the world populations are carriers of different inherited disorders of hemoglobin. Another WHO report estimates that 3% are carriers of β thalassemia and 4% are carriers of Hb-E in Bangladesh.[1] In Bangladesh more than 7000 children are born with thalassemia each year. The patients suffering of β thalassemia major and Hb-E β thalassemia do not survive more than 5 years without blood transfusion.[1] Every year near about 300,000-500,000 children are born with severe inherited disorders of hemoglobin (thalassemia and

hemoglobinopathies).[2] Out of these, 80% of them are born in middle and low income countries.[2] Hemoglobinopathies are a major health problem all over the world including India and South Asian region.[3] Worldwide, the Hb disorders are responsible for 3.4% mortality in children below 5years of age.[4] Thalassaemia results from a reduced rate of synthesis of α or β chains. B-Thalassaemia is more common in Mediterranean region while α–thalassaemia is common in Far East. Individuals with thalassemia major usually present with in the first two years of life with severe anaemia, requiring regular red blood cell transfusions.[5] Carriers of β thalassemia are clinically asymptomatic.[5] Hemoglobinopathies means structural defect of hemoglobin in which there is a substitution of single amino acid in one of their pairs of polypeptide chains. Most common hemoglobinopathies are Hb-S, Hb-C, Hb-E, Hb-D Punjab.[6] Hb–E is the most common hemoglobin variant in South–East Asia. Hb E carrier is asymptomatic. In the homozygous state of Hb–E there is mild microcytic hypochromic anaemia. HbE/β^0 thalassaemia, however resembles homozygous β^{0-} thalassaemia both clinically and haematologically.[6] Recent studies have suggested that low income status and lack of awareness are also contributing in increasing the frequency of this disease.[7] Other social factors, such as a preference to marry within the ethnic groups and consanguineous marriages; have also contributed to the increased incidence of this disease[7]. Thalassaemia carriers are today present worldwide with high frequencies in the endemic countries of Africa, the Middle East, the Indian subcontinent, Southeast Asia (Bangladesh, Thailand, Indonesia, Malaysia).[8] Management of HbEβ thalassaemia is similar to homozygous β thalassaemia. Those patients with Hb >7 gm% without complications, long term folic acid is revommended.[9] Many may benefit from Hydroxyurea therapy which decreases ineffective erythropoiesis and increases Hb with or without increase in HbF.[9] Recombinant erythropoietin alone or associated with Hydroxyurea may be useful in reducing transfusion requirements, in improving quality of life and in diminishing haemopoietic ectopic extramedullary masses.[9] However curative treatment of thalassaemia and hemoglobinopathies is bone marrow transplantion.[10] The study was carried out by history, physical examination, complete blood count, peripheral blood film, red cell indices, reticulocyte count and Hb electrophoresis (Estimation carried out by Sebia Fully Automated Capillary's -2 Flex piercing method).

Objective

1. To determine the pattern of Hemoglobinopathies and Thalassaemias in a tertiary care hospital.
2. To see the clinical profiles of the patients.
3. Genetic counseling to the patients.

Methods

This cross-sectional, observational study was conducted at the department of Haematology, Medicine and pediatrics in Rangpur Medical college, Bangladesh from January 2016 to December 2016. A total 500 consecutive diagnosed patients were included in this study. Data were collected in a preformed questionnaire. Complete blood count, red cell indices, reticulocyte counts were studied. Hb electrophoresis was performed by capillary method. Data were analyzed using SPSS version 20. Mean and + SD were calculated for numerical values and frequencies for string values.

Inclusion criteria

1. Age>one year with clinical features and abnormal haematological findings suggestive of Thalassaemia and Haemoglobinopathies.
2. Both sexes

3. Who gave consent.

Exclusion criteria

1. Age < one year
2. Incomplete data
3. Who did not give consent

Results

Total 500 cases of diagnosed Haemoglobinopathies and Thalassaemias patients were included in this study. Regarding age majority (45.2%) were >20 years age, 30.6% were between 11 -20 years, 24.2% were 1 -10 years. Regarding sex 51.2% were female and 48.8% were male. M:F ratio 1:1.04. Majority of the patients were Muslim (84%). Most of the patients were HbE/β thalassaemia (40.8%) and Hb E trait(30%). Hb E disease-24.8%, β thalassaemia trait-3.4% and β thalassaemia major-1.2%.

On clinical examination 95% patients were anaemic, 60% icteric, 43% had Splenomegaly and 19% had hepatomegaly.

Table I: Age distribution of patients

Age group	Number	%
<11	121	24.2
11-20	153	30.6
>20	226	45.2

Table II: Sex distribution of the patients

Sex	Number	%
Male	244	48.8
Female	256	51.2

Table III: Patterns of haemoglobinopathies and thalassaemic patients

Patterns of haemoglobinopathies and thalassaemic patients	Number	%
Hb Ebeta thalassaemia	204	40.8
Hb E trait	150	30.0
Hb E disease	124	24.8
Beta thalassaemia trait	17	3.4
Beta thalassaemia major	06	1.2

Table IV: Clinical manifestation of the patients

Clinical manifestation of the patients	Number	%
Anaemia	475	95
Jaundice	300	60
Splenomegaly	215	43
Hepatomegaly	95	19

Table V: Religious distribution of the patients

Religion	Number	%
Muslim	420	84
Hindu	80	16

Discussion

Thalassaemias are inherited disorders characterized by abnormal production of hemoglobin and associated with low hemoglobin production and excessive destruction of the red cells. The wide prevalence of thalassaemias and hemoglobinopathies has been attributed to migration of people from one region to another and marriages between different communities. With increasing awareness, detection of this disorder occurs during premarital screening. Hb E is the most common variant hemoglobin with a mutation in beta globin gene. Hb E trait may be co inherited with β^0thalassaemia or β^+thalassaemia. The compound heterozygous state is quite common in throughout a large part of Southeast Asia including Bangladesh with prevalence rate 30 -40%[9].

In this study, we had collected 500 cases of hemoglobinopathies and thalassaemia patients. Majority (45.2%) of the patients were >20 years age, 30.6% were between 11 -20 years, 24.2% were 1 -10 years which was almost similar to the study done by Palit S, Robiul H. Dash R who showed 35.8% > 20 years' age, 30.2% between 11 -20 years' age and 34% patients 1 -10 years' age.[1]

Regarding sex female (51.2%) were more affected than male (48.8%) which was reverse

to the study done by Rakholia R, Chaturvedi P who showed male (51.8%) more than female (48.2%).[10]

In our study 84% patients were Muslim and 16% Hindu which was reverse to the study done by Shah S J, Patel A B, Sharma VY who showed Hindu (80%) and Muslim (17.1%).[11]

Most of the patients were Hb E β thalassaemia (40.8%) which was lower than the study done by Palit S, Robiul H, Dash R who shoed (77.4%) but higher than the study done by Mondal S K, Dasgupta S, Das N who sowed (1.56%).[1,4]

In our study, Hb E Trait were (30%) which was higher than the study done by Mohanty R.B, Patel R.Z, Ross C who showed (23.9%) and Naskar S, Biswas G, Adhikari A who showed (9.02%).[12,13]

In this study Hb E disease were (24.8%) which was higher than the study done by [4]Mondal S K, Dasgupta S, Das N who showed (0.39%) and [3]Mandal P K, Maji S K, Dolai T K who showed (0.05%).

Beta thalassaemia trait were(3.4%) in our study which was lower than the study done by Hassan I Y, Khalid J.K, Helmi F who showed (22.4%) , Fabryova V, Babusik P, Sakalova A who showed (21.9%) and Bhanvadia V, Mehta D, Seikh S S who showed (15.59%).[14,15,16]

In our study, β Thalassaemia Major were 1.2% which was similar to the study done by Mondal S K, Dasgupta S, Das N[4] who showed 1.26% but lower than the study done by Palit S, Robiul H, Dash R who showed 7.5%.[1]

On clinical examination, our study showed 95% patients were anaemic which were lower than the study done by Shah S. J, Patel A. B, Sharma V. Y who showed 100%. Jaundice 60% which was higher than the study done by Shah S.J, Patel A. B, SharmaV. Y who showed 22.9%.[11] Splenomegaly 43% which was lower than the study done by Shah S.J, PatelA. B, Sharma V. Y who showed 88.6%. Hepatomegaly 19% which was lower than the study done by Shah SJ, Patel A.B, Sharma VY who showed 71.4%.

Conclusion

Thalassaemias are the most common genetic blood disease in the world. Present cross sectional observational study clearly showed majority of the patients were Hb E β thalassaemia (40.8%) and Hb E trait (30%). Females were more affected than male. Muslims were more frequently affected. Majority patients (45.2%) were >20 years age. Most of the patients had anaemia, jaundice and Splenomegaly. Prevalence of Hb E β thalassaemia and Hb E trait is sparingly alarming stage in tertiary care hospital, Rangpur. Premarital and antenatal screenings are important measures to prevent birth of children with severe Hb disorders.

Recommendation

Identifying the prevalence and pattern of haemoglobinopathies and thalassaemia is an important step in the elimination of overall burden of the disease. In this study as work was carried out in a very short and limited period of time with a small number of cases it is envisaged that a long term comprehensive study should be done in this line. The programme of prevention through carrier screening and prenatal diagnosis should receive the highest priority in future, in order to reduce drastically the birth of affected children.

References

1. Palit S, Robiul H, Das R. A study of prevalence of thalassaemia and its correlations with liver function test in different age and sex group in the Chittagong district of Bangladesh. Journal

of Basic and Clinical Pharmacy, 2012;3(4):352-356.
2. Thakur S, Sharma R, Raw S N. Incidence of Thalassaemia and Sickle Cell Disease in Chhattisgarh, Central India: Using Hardy – Weinberg Equations. J Mol Genet Med, an open access journal, 2015; 9(1):1-5
3. Mandal P K, Maji S K, Dolai T K. Present scenario of hemoglobinopathoes in West Bengal, India: An analysis of a large population. International Journal of Medicine and Public health, 2014; 4(4):496-499
4. Mandal S K, Dasgupta S, Das N. Spectrum of thalassaemias and haemoglobinopathies in West Bengal: A study of 90,210 cases by cat ion exchange high-performance liquid chromatography method over a period of 8 years. Journal of Applied Haematology, 2014; 5(3):91-95.
5. Grow K, Abrol P, Yadav R. Beta Thalassaemia in India: Current status and the challenges ahead. International Journal of Pharmacy and Pharmaceutical Sciences, 2014; 6(4): 28-33.
6. Hoffbrand A.V, Moss P.A.H. Genetic disorders of haemoglobin. Essential Haematology, 6th edition. 2011, p 91 -105
7. Ain Q U, Hassan M, Jabeen F. Prevalence of β thalassaemic Patients Associated with Consanguinity and Anti –HCV –Antibody Positivity –A Cross sectional study. Pakistan J. Zool. 2011; vol.43(1),pp.29 -36.
8. Khodaei G H, Farbod N, Saeidi M. Frequency of Thalassaemia in Iran and Khorasan Razavi. International Journal of Pediatrics, 2013; 1(1):45-49.
9. Mujawar Q,Ukkali S, Thobbi A N. Haemoglobin E β+ Thalassaemia. A case report from Bijapur, South India. Al Ameen J Med Sci, 2009; 2(1):82 -84.
10. Rakholia R, Chaturvedi P. Prevalence of β thalasaemia carrier state in Sindhi community of Wardha and evaluation of risk factors for β thalassaemia trait. Nigerian Journal of Clinical Practice .2013; 16(3):375 -380.
11. Shah S.J, Sharma V. Y: A Profile of cases of hemoglobinopathies at a Medical College. National Journal of Medical Research, 2012; 2(2):137 -139.
12. Mohanty R.B, Patel R.Z, Ross C. Prevalence of β – thalassaemia and other haemoglobinopathies in six cities in India: a multicentre study. J Community Genet, 2013; 4:33 -42.
13. Naskar S, Biswas G, Adhikari A. Prebalence of Thalassaemia and Other Hemoglobinopathies in a Northern District of West Bengal, India. IOSR Journal of Dental and Medical Sciences.2015; 14(4):43 -45.
14. Hasan I, Khalid J, Helmi F. Thalassaemia genes in Baghdad, Iraq. Eastern Mediterranean Health Journal. 1996; 2(2): 315- 319
15. Fabryova V, Babusik P, Sakalova A. NINETEEN YEARS STUDY OF BETA – THALASSAEMIA IN SLOVAKIA. Cent Eur J Public Health. 2012; 20(4):239 -243.
16. Bhanvadia V M, Mehta D, Seikh S. S. Study of prevalence of hemoglobinopathy in Sindhi community of Jamnagar, Gujrat: A pilot study. International Journal of Medicine and Public Health. 2015; 5(4):349 -352.