ORIGINAL RESEARCH PAPER

Pathology

A STUDY ON OCCURRENCE OF THALASSEMIA AND HEMOGLOBINOPATHIES IN ANTENATAL, PREMARITAL AND POST MARTIAL SCREENING IN THE DISTRICT OF MALDA, WEST BENGAL

KEY WORDS: HPLC, Malda, screening, thalassemia, Bengal

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ABSTRACT

Background: Thalassemia and other hemoglobinopathies continue to be a major public health problem in the state of West Bengal. There is significant regional variation of prevalence of hemoglobin disorders in different regions.

Aims & Objectives: The present study was conducted to find the occurrence of different types of hemoglobinopathies amongst the people undergoing screening in the district of Malda with a special emphasis on the relative occurrence in different ethnic populations.

Methodology: This research work was conducted at the thalassemia clinic and hematology laboratory of Malda Medical College and Hospital among antenatal mothers (antenatal screening), married couples (post marital screening) and children and young unmarried people (premarital screening) of Malda district. Hemoglobin analysis was done by High Performance Liquid Chromatography (HPLC).

Results: Overall, 8.7 % participants were found to be having hemoglobin abnormalities. Hemoglobin E trait was the commonest hemoglobinopathy identified, comprising 5.5% of the cases. This was followed by Thalassemia trait (2.4%), HbE homozygous (0.3%), HbS trait (0.3%), Hemoglobin E (0.1%) and hemoglobin ES (0.1%) compound heterozygotes.

Conclusion: This data reflects the importance of complete screening of the population for thalassemia and hemoglobinopathies to prevent birth of diseased children by genetic counseling.

INTRODUCTION

Thalassemia and other hemoglobinopathies continue to be a major public health problem in the state of West Bengal. These diseases can be prevented by population screening and offering genetic counseling. It is estimated there are over 25 million carriers of thalassemia and hemoglobinopathies in India.(1) The estimated prevalence of thalassemia and hemoglobinopathies in India is 3-8%. (2) There is significant variation in prevalence and relative frequency of different hemoglobinopathies across different regions of India. (3) This study attempts to estimate prevalence of thalassemia and hemoglobinopathies from the subjects screened at thalassemia clinic at Malda Medical College, Malda, West Bengal.

MATERIALS & METHODS

This study was conducted among antenatal mothers, newly married couples (post marital screening), children and young unmarried people (premarital screening) of people living in Malda district.

This study was a cross-sectional study conducted at the Thalassemia Clinic and the Department of Pathology of Malda Medical College and Hospital during the period January 2015 to June 2015. After informed consent, basic demographic details and relevant history were obtained by means of a pre-tested questionnaire form and 2 ml EDTA blood was collected in a vial and sent for testing the thalassemia laboratory. The blood samples were run in SYSMEX-KX-21 Hematology Analyzer (Sysmex Corporation) for routine hematological parameters and Variant Hemoglobin Testing System (Variant II Beta Thalassemia Short Program, Bio-Rad Laboratories) hemoglobin abnormalities by High Performance Liquid Chromatography (HPLC).

The data were finally put together for analysis.

RESULT

A total of 1017 people were screened in this study with age range of 12 to 47 years (median 18 years). 88 persons (8.7%) showed some form of abnormality in hemoglobin analysis by HPLC. Their age range was 14 to 39 years (median 18 years) and majority (53 i.e. 60.2%) were females.

530 persons underwent premarital screening for hemoglobin disorders whereas 15 persons chose to screen for the same after marriage (post-marital screening). 472 antenatal mothers also underwent the screening procedure for hemoglobin disorders. The age and sex distribution of the subjects are depicted in Tables 1, 2 & 3.

Table 1A &1B: Premarital screening

PREMARITAL all participants (530 persons)				
AGE (yrs)		**SEX**	NO	%
RANGE	12.0-32.0	M	377	71.1
MEAN	16.6 (2.3)	F	153	28.9
MEDIAN	16			

PREMARITAL abnormal findings (46 persons)				
AGE (yrs)		**SEX**	NO	%
RANGE	14-29	M	35	76.1
MEAN	17.2 (2.9)	F	11	23.9
MEDIAN	17			

Table 2: Antenatal screening

ANTENATAL			
ALL (472)		ABNORMAL (40)	
AGE		**AGE**	
RANGE	17.0-39.0	RANGE	18.0-39.0
MEAN	22.0 (3.9)	MEAN	22.1 (4.8)
MEDIAN	21	MEDIAN	20

Table 3A & 3B: Post marital screening

POST MARITAL all participants (15 persons)				
AGE (yrs)		**SEX**	NO	%
RANGE	18.0-47.0	M	6	40.0
MEAN	15 (8.4)	F	9	60.0
MEDIAN	26			

POST MARITAL abnormal findings (02 persons)				
AGE (yrs)		**SEX**	NO	%
RANGE	18.0-20.0	M	0	0.0
MEAN	19	F	2	100.0
MEDIAN	19			

Table 4 depicts the relative proportions of different hemoglobin disorders in all the screening population as well as groups like antenatal mothers, premarital and post-marital screening.

Table 4: Result of hemoglobin analysis in different screening groups

Hemoglobin Characteristic	Antenatal Screening	Postmarital Screening	Premarital Screening	Grand Total	
Beta thal trait	17		7	24	2.36%
E Homozygous	1	1	1	3	0.29 %
E trait	19	1	36	56	5.51%
EB	1			1	0.10%

ES			1	1	0.10%
S trait	2		1	3	0.29%
Normal analysis	432	13	484	929	91.35%
Grand Total	**472**	**15**	**530**	**1017**	
All Abnormal Together	**88 (8.7%)**				

It is evident that about 8.7% people undergoing screening had some form of hemoglobin disorders albeit almost all of them are carriers. HbE (5.5%) was the most common hemoglobin abnormality followed by beta thalassemia trait (2.4%). Screening also detected presence of HbE homozygous (0.3%), E-beta thalassemia (0.1%), HbES compound heterozygote (0.1%) and HbS trait (0.3%).

Table 5: Religion of all participants

	All participants		Abnormal hemoglobins detected in	
Religion	**No. of participants**	**%**	**No. of participants**	**%**
CHRISTIAN	2	0.2	0	0
HINDU	641	63.0	49	55.7
MUSLIM	374	36.8	39	44.3
Grand Total	**1017**	**100.00**	**88**	**100.0**

Table 6: Proportion of abnormal hemoglobins in different religion groups

GROUP	Religion		%
ANTENATAL SCREENING	HINDU	19	47.5
	MUSLIM	21	52.5
		40	**100.0**
POSTMARITAL SCREENING	HINDU	1	50.0
	MUSLIM	1	50.0
		2	**100.0**
PREMARITAL SCREENING	HINDU	29	63.0
	MUSLIM	17	37.0
		46	**100.0**

When we see the religion-wise break up of participants (vide table 5) it was found that 63% participants were Hindu, 36.8% were Muslim and 0.2% were Christian. In the subjects detected to have abnormal hemoglobins, 55.7% were Hindu and rests were Muslim. From Table 6 it is evident that hemoglobin abnormalities were found more often in Muslim antenatal mothers than their Hindu counterparts while in premarital screening, more number of Hindus were detected with abnormalities than Muslims.

Table 7: Caste-wise break-up of participants with abnormal hemoglobin analysis

Religion	Caste	Abnormal	
		No.	% of Participants of same category
Christian	GEN	0	0.0
	ST	0	0.0
		0	0.0
Hindu	GEN	14	6.7
	OBC	5	8.1
	RAJBANSHI	7	29.2
	SC	22	8.4
	ST		0.0
	UNKNOWN	1	1.3
		49	7.6
Muslim	GEN	29	15.8
	OBC	5	9.8
	UNKNOWN	5	3.6
		39	10.4
Grand Total		88	8.7

From table 7 we see that most caste categories reflect the proportion of overall participants in the share of abnormal hemoglobin variants except the Rajbanshi community which shows a high occurrence (29.2%). Muslim general caste category with 15 % occurrence is also quite above the overall proportion of 8.7%.

Table 8: Hemoglobin analysis amongst Rajbanshis

Diagnosis	No.	%
Beta thal trait	1	4.2
E Homozygous	1	4.2
E trait	5	20.8
Normal	17	70.8
TOTAL	**24**	**100.0**

Rajbanshi people showed mostly HbE trait as abnormality apart from single cases of HbE disease and Beta thalassemia trait each.

Hematological parameters of the persons with hemoglobin abnormalities reveal presence of variable degrees of microcytic hypochromic anemia with anisocytosis. (Table 9)

Table 9: Hematological parameters of persons with hemoglobin abnormalities

	Mean	Standard Deviation	Range
RBC(million/cumm)	4.92	0.82	2.39-6.71
Hb(gm/dL)	11.41	2.04	5.3-15.9
PCV (l/l)	35.52	5.93	19.5-49.1
MCV (fl)	72.65	7.85	51.6-89.7
MCH (pg)	23.76	3.32	16.9-33.9
MCHC (gm/dL)	31.94	3.14	21.4-41.7
RDW (CV%)	16.3	2.66	12.6-31

DISCUSSION

This study reveals 8.7% of screened individuals had hemoglobin abnormalities which is a little lower than the previous studies done in West Bengal (4) (5). In our study HbE trait was most commonly found in the screened individuals. Our finding is similar to the findings of a study done by Goswami B K, Pramanik R, Chakrabarty S et al in North Bengal Medical College, Siliguri, West Bengal. (6) On the other hand, in a study done by Mandal P K et al with sampling done from all over West Bengal, Beta thalassemia trait was found to be the most common abnormality (6.61%) followed by HbE trait (2.68%). Studies done in Burdwan and Bankura districts of West Bengal by Jain B B et al and Mondal Bikash et al respectively also found Beta Thalassemia trait as the most common hemoglobin abnormality. (7) (8)

Although it was a screening based study where diseased cases naturally get excluded, a single case of an antenatal mother with E Beta thalassemia was found during the study further attesting the vast spectrum of clinical severity of E Beta thalassemia. (9)

We do not get religion and caste based break-ups of thalassemia carriers and patients in previous studies so we made an attempt to identify ethnic focus of hemoglobin abnormality if present.

It was seen that majority of people choosing premarital counseling were Hindu and majority of antenatal mothers undergoing screening are Muslim. However the difference in proportions was not statistically significant.

Despite only a small number of participants, we calculated specifically the proportion of people with abnormal hemoglobin in Rajbanshi community, a separate ethnic community in North Bengal, which is known for a high prevalence of hemoglobinopathies with very high proportion HbE trait. (6)

Apart from the Rajbanshis other caste categories do not show any significant difference in the proportion of participants with abnormal hemoglobins.

CONCLUSION

Our study attempted to measure the burden of hemoglobin disorders on the society based on screening. It showed about 8.7% occurrence of hemoglobinopathies in screening population which very well corroborates with other population based studies. It also reiterates the high prevalence of HbE in North Bengal compared to South Bengal. It also attests to the fact that Rajbanshi

community has a high prevalence of hemoglobin disorders, especially HbE disorders compared to other hemoglobinopathies. So the message is to cover everyone in every corner of the nation under the thalassemia screening program so that birth of diseased children can be prevented by genetic counseling.

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