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Original Research Article

Screening for hemoglobinopathies in a socially disadvantaged population from a rural district of West Bengal, India

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ABSTRACT

Background: Detection of traits/carriers plays an important role in preventing the birth of a thalassemic child. West Bengal, one of the eastern states in India is the home to a bulk of socially challenged population including scheduled castes and scheduled tribes among others. The present study aimed to detect the prevalence of different hemoglobinopathies in a socially challenged district of West Bengal.
Methods: In this retrospective cross sectional study thalassemia detection camps were organized at the community level over a period of four years. Venous blood samples were subjected to complete hemogram and high performance liquid chromatography (HPLC). In few difficult cases samples were sent to the reference laboratory for molecular characterization. The prevalence of heterozygous, homozygous or compound heterozygous states of different thalassemias and hemoglobinopathies across various respondent groups (e.g. children, premarital, postmarital and antenatal) and existing caste categories (scheduled tribes, scheduled caste and general) were analyzed.
Results: We analyzed a total of 114,606 HPLC reports; 18681 (16.30%), 15438 (13.47%) and 80487 (70.23%) cases belonged to scheduled tribes, scheduled castes and general category respectively. Out of 114,606 cases, 11,001 (9.6%) had revealed abnormal hemoglobins; beta thalassaemia trait was the most common (6.63%; n=7602) across all subgroup analysis. Among others, HbE trait, sickle cell trait and HbD trait were detected in 1788 (1.56%), 1362 (1.18%) and 126 (0.11%) cases respectively.
Conclusions: Beta thalassaemia trait and HbE trait are the common haemoglobin variants in this rural district of West Bengal. The prevalence of sickle gene revealed in the present study is much less than previous studies in the locality.

Keywords: West Bengal, Jhargram district, Socially challenged population, Thalassemia screening

INTRODUCTION

West Bengal, one of the eastern states in India is the home to 9.13 crore population (as per census 2011), has a relatively higher burden of thalassemia; estimated 6-10% of the population are carriers of the disease.[1-3] West Bengal is the first state in India to launch the State Thalassaemia Control Programme (STCP) in 2008 for prevention of thalassemia.[4] Beta-thalassaemia, E-beta thalassaemia, Sickle cell anemia, and hemoglobin D Punjab are the common hemoglobinopathies in India including West Bengal.[5,6] The various groups identified for social disadvantaged status include- scheduled castes (SC), scheduled tribes (ST), backward class (BC), women and minorities.[7] In constitutional terms, dalits are generally known as the scheduled castes; they suffer discrimination in education, health care and equal

treatment before the law.[7,8] In India, population of the scheduled castes constitute 16.2% of the total population; Uttar Pradesh has the largest scheduled caste population followed by West Bengal and Bihar. India has many tribal communities; they are one of the most vulnerable sectors of the population exposed to many health hazards as their residence is usually located in isolated places, embrace rigid customs, high illiteracy and separation from modern amenities.[9] There is a variety of tribes found in almost all the nooks and corners in the state of West Bengal; about 38 in number, the tribes comprise nearly 2% of the entire population. Generally confined to the rural belt of the state; a small portion of them has now moved to the urban belt, in search for employment and a better lifestyle.[10] More than half of the total ST population of the state are concentrated in four districts, namely (undivided) Medinipur, Jalpaiguri, Purulia, and Bardhaman.[11] The said (undivided) Medinipore district was subsequently divided into three independent districts namely East Midnapore, West Misnapore and Jhargram Districts (the last one formed on 4th April, 2017). As per 2011 census, Jhargram district had a population of 1,136,548, of which 96.52% resides in the rural areas; 20.11% of the total population belonged to scheduled castes and 29.37% belonged to scheduled tribes with high residence of certain tribal ethnic groups e.g. Oraon and Munda.[12] Previous studies published in the years 2012 and 2014 respectively by Dolai et al and Mandal et al from the district of (undivided) West Midnapore (also included the data from the then Jhargram sub-division) detected abnormal hemoglobins (Hbs) in 16.23% and 11.62% of total HPLC done in the district.[6,13] But, no separate data available in the published literature about the prevalence of different hemoglobinopathies in this rural district of West Bengal with a socially disadvantaged population of around 50%. The present work was performed to assess the prevalence of different thalassaemias and hemoglobinopathies diagnosed by HPLC on screening programme at the community level in this district with high number of socially challenged population.

METHODS

The retrospective study was conducted over a period of 4 years from June, 2016 to May, 2020 by thalassaemia detection centres at district headquarters of Jhargram District. Thalassemia carrier detection camps were organized by Paschim Medinipur Thalassaemia Prevention Society (PMTPS) mostly in schools, colleges and clubs in the pockets of rural areas and also included antenatal mothers attending different hospitals for antenatal checkup. Informed and written consent were taken in all cases before blood sampling. A total of 114,606 individuals volunteered for the test. Whole blood sample was collected in the tri-potassium EDTA vacuum container from each subject and transported to the laboratory within 3-5 hours of collection. Complete hemogram was done by automated blood cell counter (Sysmex KX-21; Sysmex Corporation, Kobe, Japan), then subjected to high performance liquid chromatography (HPLC) by VARIANT-IITM (Bio-Rad Laboratories, Hercules, CA, USA) for estimation of haemoglobin fractions. In some difficult cases, where accurate interpretation was not possible by HPLC, samples were sent to the reference laboratory at Nilratan Sircar Medical College, Kolkata for molecular characterization and confirmation of diagnosis. Samples from parents, siblings and close relatives also tested as part of extended family screening. Simple Statistical analysis was done with help of Microsoft Office Excel 2007.

RESULTS

In this retrospective analysis, a total of 114,606 samples were collected and analysed. Overall, a total of 11,001 (9.6%) cases had revealed abnormal Hbs (Table 1).

Table 1: demographic profile of patients and HPLC findings (n=114,606).

Caste/category	Respondent group	Age (years), Median (range)	Sex, Male/female; Ratio (n/n)	Total HPLC	Normal Hb, N	Abnormal Hb, N (%)
Scheduled tribes	Children	3 (1-15)	1.19 (276/231)	507	423	84 (16.56)
	Premarital	16 (11-41)	0.56 (3465/6147)	9612	8772	840 (8.74)
	Post marital	25 (12-54)	2.39 (3810/1596)	5406	4929	477 (8.83)
	Antenatal mother	21 (14-38)	0/3156	3156	2847	309 (9.79)
	Total	17 (1-54)	0.68 (7551/11130)	18681	16971	1710 (9.15)
Scheduled caste	Children	4 (1-12)	1.05 (192/183)	375	297	78 (20.8)
	Premarital	14 (11-36)	0.52 (3264/6213)	9477	8481	996 (10.51)
	Post marital	25 (13-69)	2.39 (2391/999)	3390	3003	387 (11.41)
	Antenatal mother	14 (11-36)	0/2196	2196	1926	270 (12.3)
	Total	18 (1-69)	0.61 (5847/9591)	15438	13707	1731 (11.21)
General category	Children	3 (1-16)	0.87 (552/483)	1035	867	168 (16.23)
	Premarital	16 (13-45)	0.61 (24606/40524)	65130	59253	5877 (9.02)
	Post marital	27 (15-65)	0.38 (2493/6570)	9063	8028	1035 (11.42)
	Antenatal mother	21 (14-41)	0/5259	5259	4779	480 (9.13)
	Total	16 (1-65)	0.52 (27651/52836)	80487	72927	7560 (9.39)
Grand total	-	17 (1-69)	0.59 (41,049/73,557)	114,606	103,605	11,001 (9.6)

Table 2: Detail account of abnormal haemoglobins (n=11,001) across various respondent groups and caste/category.

Respondent group	Caste /category	Total HPLC	Abnormal Hb, n (%)	Beta thalassemia trait	HbE trait	Hb D trait	Hb S trait	HPFH	Beta thalassemia major	HbE beta thalassemia	Hb EE disease	HbS beta thalassemia	Sickle cell disease
Children	Scheduled tribes	507	84(16.56%)	66	6	0	6	3	3	0	0	0	0
	Scheduled caste	375	78 (20.8%)	51	6	0	21	0	0	0	0	0	0
	General category	1035	168 (16.23%)	120	15	0	24	3	0	6	0	0	0
	Total	1917	330 (17.21%)	237 (12.36%)	27 (1.41%)	0	51 (2.66%)	6 (0.31%)	3 (0.15%)	6 (0.31%)	0	0	0
Premarital	Scheduled tribes	9612	840 (8.74%)	738	6	3	90	0	0		3	0	0
	Scheduled caste	9477	996 (10.51%)	516	159	39	273	0	0	0	6	3	0
	General category	65130	5877 (9.02%)	4029	1323	69	366	12	9	9	12	12	3
	Total	84219	7713 (9.16%)	5283 (6.27%)	1488 (1.77%)	111 (0.13%)	762 (0.90%)	12 (0.01%)	9 (0.01%)	9 (0.01%)	21 (0.02%)	15 (0.017%)	3 (0.003%)
Post marital	Scheduled tribes	5406	477 (8.83%)	408	6	3	57	0	0	0	0	3	0
	Scheduled caste	3390	387 (11.41%)	213	33	3	132	3	0	0	3	0	0
	General category	9063	1035 (11.42%)	723	147	3	141	3	3	6	6	3	0
	Total	17859	1899 (10.63%)	1344 (7.52%)	186 (1.04%)	9 (0.05%)	330 (1.15%)	6 (0.03%)	3 (0.015%)	6 (0.03%)	9 (0.05%)	6 (0.03%)	0
Antenatal mother	Scheduled tribes	3156	309 (9.79%)	234	12	3	60	0	0	0	0	0	0
	Scheduled caste	2196	270 (12.3%)	141	21	3	96	0	0	0	0	2	3
	General category	5259	480 (9.13%)	363	54	0	63	0	0	0	0	0	0
	Total	10611	1059 (9.98%)	738 (6.95%)	87 (0.82%)	6 (0.056 %)	219 (2.06%)	0	0	0	0	6 (0.056%)	3 (0.028%)
Grand total	-	114,606	11,001 (9.6%)	7602 (6.63%)	1788 (1.56%)	126 (0.11%)	1362 (1.18%)	24 (0.022%)	15 (0.015%)	21 (0.016%)	30 (0.03%)	27 (0.025%)	6 (0.006%)

Out of all the abnormal Hbs, 7602 (6.63%) cases were β-thalassaemia trait, the most common thalassemia trait/carrier detected in the study (Table 2).

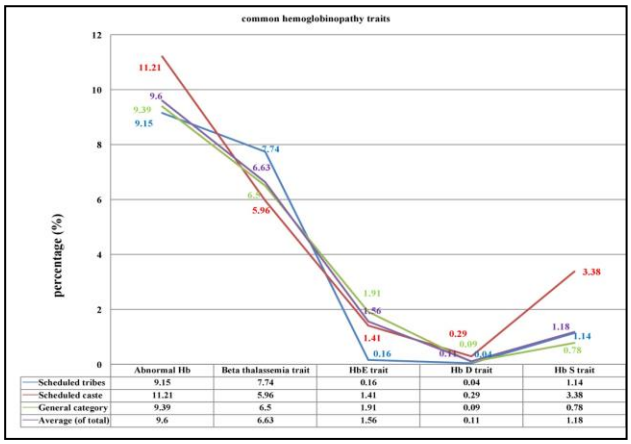

Figure 1: Details of the four common hemoglobinopathy traits/carriers detected across different castes/category.

Among others, HbE trait, sickle cell trait and HbD trait were detected in 1788 (1.56%), 1362 (1.18%) and 126 (0.11%) cases respectively. Hereditary persistence of fetal haemoglobin (HPFH) trait was detected in 24 (0.022%) cases. As revealed in the present study, among homozygous or compound heterozygous states, HbEE disease was the commonest (0.03%; n=30) followed by HbS beta thalassaemia (0.025%; n=27), HbE beta thalassaemia (0.016%; n=21) and beta thalassemia major (10.015%; n=15). The prevalence of heterozygous, homozygous or compound heterozygous states of different thalassemias and hemoglobinopathies across the respondent groups (e.g. children, premarital, postmarital and antenatal) and existing caste categories (scheduled tribes, scheduled caste and general) are detailed in Table 2. The detection rate of the common thalassemia trait/carrier detected in the study across existing caste categories are shown in Figure 1.

DISCUSSION

The most effective option for controlling the community load of thalassaemias and other hemoglobinopathies is prevention of birth of affected children by premarital screening and also screening and counselling of the young adolescents. Overall outcome of thalassemia screening and counselling depends on multiple attributes that may include place of residence (urban and rural), religion, level of education (no education versus primary/secondary/higher education), work status (not working versus paid job) and socio-economic status.[14] Jhargram district (formerly a sub-division) was formed

on 4th April, 2017, after bifurcation from the Paschim Medinipur district. As per 2011 census, Jhargram district had a population of 1,136,548, of which 96.52% reside in the rural areas. The socially disadvantaged population constitute about 50% of the total population; 20.11% of the total population belonged to scheduled castes and 29.37% belonged to scheduled tribes with high residence of certain tribal ethnic groups e.g. Oraon and Munda.[12]

As shown in Table 1, out of 114,606 cases, 18681 (16.30%), 15438 (13.47%) and 80487 (70.23%) cases belonged to scheduled tribes, scheduled castes and general category respectively. Overall, prevalence of abnormal haemoglobins was higher (11.21%) in scheduled castes followed by general category (9.39%) and scheduled tribes (9.15%). In subgroup analysis (Table 2), among different respondent groups, children had the highest rate of abnormal haemoglobins (17.21%) ranging from 16.23-20.8% in different caste/categories that reflects the poor level of consciousness among people and require intense counselling to decrease the load of thalassaemia in the community. Across all respondent groups and caste/categories, beta thalassaemia trait was the predominant heterozygous state prevalent (overall, 6.63%; range: 6.27-12.36%) in the community followed by HbE trait (overall, 1.56%; range: 0.82-1.77%). In a study in the tribal population of North-East India by Ghosh et al haemoglobin E was the prevalent trait followed by haemoglobin S.[15] But, the among the tribals working in the tea gardens of Assam by Teli et al revealed prevalence of beta thalassemia trait of 10% and 31% in the Oraon and Munda ethnic groups of tribal populations coming from West Bengal; the present study corroborates with this.[16] Studies by Dolai et al and Mandal et al from the district of (undivided) West Midnapore (that also included the data from the then Jhargram sub-division, a separate district now) has shown prevalence of beta thalassaemia trait in the community with reported detection rate of beta thalassaemia trait of 10.38% and 6.61% respectively.[6,13] Majumdar et al studied on beta thalassemia in different tribal population in West Bengal and reported prevalence of beta thalassemia over HbE trait from the southern part of Bengal.[17] Scheduled tribes had shown (Figure 1) the higher prevalence (7.74%) of beta thalassemia trait in comparison to others, whereas HbE trait was more common in the general category (1.91%). HbS came out to be the third common trait (1.18%); again it was much higher in children among all respondent groups. In the present study population (Figure 1), Hbs S trait was more common in scheduled castes (3.38%) than the scheduled tribes (1.14%) and general category (0.78%). Chattopadhyay et al from the same locality found high detection rate of Hb S (2.22%).[18] Kaur et al reported prevalence of sickle gene to be 0-18% in north eastern India and Colah RB et al reported an incidence of HbS trait of <5% in West Bengal.[19,20] But a recent study by Ray et al from Kolkata in a study in school going children from tribal populations of almost all districts of West Bengal reported HbS in only 0.4% of the study population; they postulated that the widely varying prevalence of alpha deletion (83%) and exceptionally low prevalence of HbS (0.4%) may be a consequence of epistasis.[21] The fourth common hemoglobinopathy (0.11%) in the present study was HbD; more common in scheduled castes than others (Figure 1).

Overall, among homozygous or compound heterozygous states (Table 2), HbEE disease was the commonest followed by HbS beta thalassaemia, HbE beta thalassaemia and beta thalassemia major; almost equally distributed among the respondent groups across the caste/categories. This was in contrast to many other studies from this region of the country by Mohanty et al, Mandal et al and Chattopadhyay et al where in a community with high prevalence of beta thalassemia trait and HbE traits, HbE beta thalassaemia comes out as the commonest compound heterozygous thalassemia.[5,13,18] HbS beta thalassemia, a clinically significant thalassemia was detected in 0.025% (n=27) cases in the present study was much lower than by other previous study from the locality that had shown a prevalence of 0.15%.[13]

As evident from Table 2, in spite of high prevalence of different traits especially beta thalassemia trait and HbE trait, the detection rate of different homozygous and compound heterozygous thalassaemias are much lower in the present study in comparison to many other previous studies.[6,13,18] This may be a direct effect of increasing the number of thalassemia detection camps in recent years and good level of counselling of the target population by health care personnel. Preventive programmes carried out based on detection of heterozygous states and counselling to avoid marriage between carriers and/or foetal diagnosis have been very effective in reducing birth of a thalassaemic child. Good health of the people of any particular area is not reflected by the availability of healthcare facility only but also by how the people do utilize this facility. Utilization of modern healthcare facilities is very poor among tribal population; innovative strategies to be adopted to increase its acceptance by involving the local people and educate them by the means which they can easily accept.[22]

Thus, the key features highlighted in the present study are: a) Jhargram district is a rural district (96.52% of total population resides in the rural areas) in West Bengal (one of the eastern states in India) that is the home to a bulk (50%) of socially challenged population including scheduled castes and scheduled tribes b) Among the population screened for hemoglobinopathies, 16.30%, 13.47% and 70.23% cases belonged to scheduled tribes, scheduled castes and general category respectively c) Beta thalassaemia trait and HbE trait are the common haemoglobin variants across all groups d) Revealed in the present study is much less than previous studies

And, among few limitations in the present study, the most important are: Only a small fraction (10.08%; n=114,606/1,136,548) of the total population in the

district is screened; though the scheduled tribes and scheduled castes constitute nearly 50% of total population in the district, but a small fraction of them (16.30% and 13.47% respectively) are screened for till now, reflecting a large number of these socially disadvantaged population are yet to be screened for hemoglobinopathies; across all respondent groups and different caste/category, voluntary screening of males was almost half that of females; as 96.52% of the population resides in the rural areas, more number of thalassemia detection camps to be organized in the remotest village areas.

CONCLUSION

Beta thalassaemia trait and HbE trait are the common haemoglobin variants in this rural district of West Bengal with high number of socially disadvantaged population. The prevalence of sickle gene revealed in the present study is much less than previous studies in the locality. Detection of heterozygous states and counseling to avoid marriage between carriers has been very effective in reducing birth of a thalassaemic child. The screening programme requires involvement of more subjects from the socially disadvantaged population including males to get more homogenous results.

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