Proceedings of the Indian National Science Academy (2025) 91:940–943
https://doi.org/10.1007/s43538-024-00367-x

RESEARCH PAPER

Thalassemia carrier screening among the college going students of the rural areas of West Midnapore, India

Amrita Panja[1,2] · Biplab Kahar[2] · Mantu Kumar Das[3] · Tuphan Kanti Dolai[4]

Received: 27 April 2024 / Accepted: 29 October 2024 / Published online: 3 January 2025

Abstract

Introduction Hemoglobinopathies are common genetic disorders of hemoglobin, which can be prevented by population screening and genetic counseling. India is within the thalassemia belt of the world where the carrier frequency ranges between1 and 17%.The remote areas of West Midnaporeis not so much explored till now and population screening is essential among the pre-marital age group for identification and inhibition of high-risk marriages.

Objective The present study was undertaken to find out the spectrum of hemoglobinopathies among the college going students.

Materials and methods A total of 221 college going students of Ghatal Rabindra Satabarsiki Mahavidyalaya (GRSM) were screened. Cases were diagnosed as thalassemias and hemoglobinopathies by Bio- Rad variant II HPLC system by β-thal short program. The retention times, proportion of the haemoglobin (%), and peak characteristics for all hemoglobin (Hb) fractions were documented.

Results Amongst 221 cases included in the present study and 17 (7.69%) were identified to have thalassemia trait. Out of them, 9 (4.07%) cases had β-thalassemia trait, 6 (2.71%) cases of HbE-trait. There was one subject with HbD-Punjab variant and another one was compound heterozygote for HbE and HbS hemoglobin variant.

Conclusions The screening for thalassemia should be included as a part of regular blood testing among the college going students which will provide idea about the extent of hemoglobinopathy in the rural area of West Midnapore.

Keywords Population screening · Thalassemia · Hemoglobinopathies · High performance liquid chromatography (HPLC) · West Midnapore

Introduction

Hemoglobinopathies are autosomal recessive disorders which can be characterised by either reduced or absent synthesis of one or more globin chains of hemoglobin molecule (Balgir 1996; Borgna-Pignatti and Galanello 2004; Mondal and Mandal 2016). The homozygous or compound heterozygous states for β-thalassemia exhibit variable degree of phenotypic heterogeneity; although without transfusion death usually occurs within the first few years (Weatherall and Clegg 2001). It is the commonest single gene disorders in the world which was first observed in the Mediterranean population and causing significant morbidity and mortality in India and other countries throughout the world. Thalassemia becomes the major public health problem throughout the world especially in South East Asian countries like India (Weatherall 2008; Goh et al. 2020). According to World Health Organization (WHO) 7% of the world population are the carriers for hemoglobin disorders

✉ Tuphan Kanti Dolai
tkdolai75@gmail.com

Amrita Panja
amritapanja87@gmail.com

[1] Molecular Biology and Human Genetics Laboratory, The University of Burdwan, Burdwan, West Bengal 713104, India

[2] Panchakot Mahavidyalaya, Neturia, Sarabari, Purulia, West Bengal 723121, India

[3] Ghatal Rabindra Satabarsiki Mahavidyalaya, West Midnapore, Ghatal, West Bengal 721212, India

[4] Department of Haematology, Nilratan Sircar Medical College and Hospital, Kolkata, West Bengal 700014, India

(Modell and Darlison 2008). The prevalence of thalassemia and other hemoglobinopathies varies according to the geographical locations.

In India, 0.37 per 1000 fetuses have Hb disorder (Borgna-Pignatti et al. 2009) and in certain population, the mutation rate for thalassemia is as high as 17% (Mondal and Mandal 2016). It is estimated that there are over 25 million carriers within the country (Saxena and Phadke 2002). The frequency of thalassemia trait in India is 3–8% (Verma et al. 1992; Parthasarathy 2012). It was documented that yearly 10,000 children with β-thalassemia major are born in this country which comprises 10% of the total number in the world (Varawalla et al. 1991). HbS, HbD, HbLepore, HbJ and SD are the different types of hemoglobin variants found in different states throughout the country (Kamble and Chatruvedi 2000).

According to 2011 census in India, the population of West Bengal was around 9.13 crore which encompasses around 7.6% of the total population (Demographic Features: Public Health in West Bengal—Current Status and Ongoing Interventions. xxxx). It has been found that around 6–10% of the population is the carriers for thalassemia. According to the published reports, the carrier rate for β- thalasssemia and HbE were 3.1% and 4.1% respectively (Tyagi et al. 2003). The frequency (22%) of HbE is very high in Kolkata and HbE/β-thalassemia is regarded as the predominant hemoglobinopathy in this part of the country (Dolai et al. 2012). In a recent study, the carrier frequency in rural West Bengal is reported to be around 16.23% in general Bengali population (Shrivastav et al. 2014). Although there is scarcity of proper reports obtained from urban areas of West Midnapore.

The present work was conducted to assess the prevalence rate of different hemoglobin variants by generating awareness and successful screening programme among the college going students for carrier detection which is essential to recognize asymptomatic carriers who have an increased risk of having a child with thalassemia. The college going students from the part of the next generation and their screening would be helpful to the society. Till now, there is no proper data available from rural areas of West Midnapore which is situated in the eastern zone of India. This will be the first epidemiological study from countryside area of Ghatal district, West Midnapore. As the proper data regarding the prevalence of hemoglobinopathies in this region is scarce, it is important to assess the extent of burden for hemoglobinopathies in this rural region surrounding Ghatal, West Midnapore.

Materials and methods

Subjects

Ethical permission and study design

Prior to the initiation of the screening programme, ethical clearance from Institutional Ethical Committee (IEC) was obtained from each of the participants before including them as the study population. The work was conducted in compliance with the declaration of Helsinki. Informed consent has been obtained from all the subjects who participated in this study. A questionnaire was framed covering the important points of clinical history of illness, blood transfusion and family background.

Sample collection

About 3 ml of blood sample was collected in tri-potassium EDTA vacuum container. Collected samples were transported under cool conditions to the laboratory within 3–6 h of collection and stored at 2–8 °C. The hematological profiling and identification of hemoglobinopathies were done in Kharagpur SD Hospital, West Midnapore.

Diagnosis of hemoglobinopathies

The diagnosis of thalassemia and different types of hemoglobinopathies is based on hematological profile, clinical history, and results obtained from high performance liquid chromatography (HPLC). For determining the red cell indices the blood samples were tested by automated blood cell counter Beckman Coulter LH 780 (Beckman Coulter, Inc., Brea, CA, USA). Hemoglobin (Hb), red blood cell count (RBC), mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH), mean corpuscular hemoglobin concentration (MCHC), hematocrit and red cell distribution were examined. Peripheral smear was observed to look for microcytic hypochromic RBCs and target cells for diagnosis of any hematological abnormalities.

Hemoglobin variants and thalassemia were identified by using VARIANT TM (Bio-Rad Laboratories, Hercules, CA, USA). HbA2 value of 3.3 to 3.8% was regarded as borderline and the value of over 3.9% was considered as β-thalassemia trait (Tyagi et al. 2003; Dolai et al. 2012; Shrivastav et al. 2014). The studied samples having mean corpuscular volume (MCV) ≤76 fl and mean corpuscular hemoglobin (MCH) ≤27 pg were further investigated (Kumar et al. 2015).

Results

A total of 221 cases (male: 44, female: 177) were studied. Of these, 200 (90.49%) had normal and 21 (9.5%) showed abnormal hemoglobin fraction on HPLC. Out of 21 cases of traits; 9 (4.07%) cases had shown β-thalassemia trait, 7 cases (3.16%) HbE trait, 1 (0.45%) case with heterozygous for HbE and HbS, 1 with (0.45%) HbD-Punjab trait, 3 cases (1.35%) had shown slightly elevated level of fetal hemoglobin traits. The hematological profile of the subjects with different types of hemoglobin variants has been described in Table 1. The frequency of β-thalassemia trait is 55.56% in Bengali Hindus, 33.33% in Muslims, followed by Santhal (11.11%) whereas HbE is comparatively higher in Rajbanshis (57.14%), and followed by Muslim (28.57%) as well as Bengali Hindu (14.28%). In the present study only one student from Marwari community possessed HbD variant whereas HbS has been obtained from one Santhali individual.Six subjects (2.71%; 5 female, 1 male) had shown lower level of hemoglobin (8.06 ± 0.86) and suggested for serum ferritin testing to assess the iron load in body. Anemia is a major health issue especially in developing countries. It is commonly observed among people at any stage of life although among the college going teenage girls and women with child bearing age group. Three subjects had mildly elevated HbF level (3.2± 0.44). One subject whose report was regarded as inconclusive (HbA2 3.7%, HbF 0.7%, Hb 13.9 g/dl), suggested for repetition of HPLC.

Discussion

Hemoglobinopathies are one of the major health issues throughout the world including India (Weatherall 2011). Previously, one of the studies done among the rural areas of West Bengal revealed the frequency of abnormal hemoglobin fraction is 11.62% after testing with HPLC (Mandal et al. 2014). Another study done among the Bengali antenatal mother and premarital group showed the frequencies of hemoglobinopathies are 9.09% and 10% respectively (Jain et al. 2012). One more study done among the general population in different rural areas of Eastern India showed the carrier rate is 13% (Basak et al. 2010). Therefore, current study shows the frequency is in between the previous studies done among the Bengali population. The findings of the present study are unique, as this population has not been studied till now regarding thalassemia screening. Bengali population is the admixture of several communities due to high rate of migration at different time points. Therefore, thalassemia screening in regional basis is very essential. The studied area, Ghatal is in the district West Midnapore which is situated at the western part of the state West Bengal, India. In this area several tribal communities and various castes exist adjacently which indicates high rate of genetic admixture. The prevalence of mutant alleles for hemoglobinopathies is quite different from other ethnic population residing in different states of India. With the best of our knowledge, it is the pioneer thalassemia screening programme done among the undergraduates of Ghatal Rabindra Satabarsiki Mahavidyalaya where students use to come from different villages situated nearby the urban area of Ghatal. Several tribal communities including Santals, Bhumij, Mundas, Lodhas, Kodas, andMahalis along with Hindu, Muslim, Sikh, and other general castes inhabit in the surrounding areas of Ghatal, West Midnapore who have participated in the present study. The diversities in the genetic constituents of the studied population make them distinctive from the population of other districts of West Bengal. Proper awareness regarding the medical, social and financial burden of the disease should be generated among the common people to reduce the social stigma of thalassemia (Bain 2006). The present study conducted among the students coming from rural areas of West Midnapore, undoubtly useful to reveal the existence of different types of hemoglobinopathies particularly in this geographical belt. Beta thalassemia is the most common hemoglobinopathy in the Bengali population followed by HbE. The present study is primarily a diagnostic screening test and a few cases are inconclusive or silent type which require further investigations like DNA analysis for mutation test and iron study. The identification of the existence of different types of hemoglobin variants in heterozygous condition is important as this may cause in the formation of lethal hemoglobin diseases in homozygous condition.

Table 1. Ethnicity wise distribution of different heterozygotes for hemoglobin variants in the present study (n = 221)

Type	Ethnic groups					Number	Percentage (%)
	Bengali Hindu	Muslim	Santals	Rajbanshis	Marwari		
β-thalassemia trait	5	3	1	–	–	9	4.07
HbE trait	1	2	–	4	–	7	3.16
HbE+HbS	–	–	1	–	–	1	0.45
HbD-Punjab	–	–	–	–	1	1	0.45
Mildly elevated HbF	2	1	–	–	–	3	1.35

Large scale carrier screening and prenatal diagnosis should be included in a regular schedule for preventing high risk marriages and thus reducing the birth of affected children. The management and control of hemoglobinopathies is the utmost requirement for the health workers, policy makers, medical and health care providers. Moreover, mass awareness, and genetic counseling are still the most important requisite.

Acknowledgements The authors are thankful to all the members of the Thalassaemia Society of Midnapore District, Paschim Medinipore (West Bengal, India). We are grateful to all the technical staff and medical laboratory Technologist of Kharagpur SD hospital, Kharagpur, West Midnapore for providing the necessary documents and methodological help.

Funding Nil.

Declarations

Conflict of interest The authors declared no conflicts of interest.

References

Bain, B.J.: Other significant hemoglobinopathies. In: Hemoglobinopathy diagnosis, vol. 367 2/e, Blackwell Publishing. 191–213 (2006)

Balgir, R.S.: Genetic epidemiology of the three predominant abnormal hemoglobins in India. J. Assoc. Phys. India **44**(1), 25–28 (1996)

Basak, J., Majumdar, D., Mukhopadhyay, S., Dasgupta, S., Das, P., Pal, N., Dey, S.: Population screening for Thalassemia in West bengal with a goal to make zero Thalassmia. Blood **116**(21), 5175 (2010)

Borgna-Pignatti, C., Galanello, R.: Thalassemias and related disorders: Quantitative disorders of hemoglobin synthesis. In: Greer JP, Foerster J, Lukens JN, Rodgers GM, Parakevas F, Glader B, editors.Wintrobes Clinical Hematology. 11th ed. Philadelphia: Lippinco Ĵ Willams and Wilkins. 1319- 65 (2004)

Borgna-Pignatti, C., Galanello, R.: Thalassemias and related disorders: Quantitative disorders of hemoglobin synthesis. In: Greer, J.P., Foerster, J., Rodger, G.M., Paraskevas, F., Glader, B., Means, R.T. (eds.) Wintrobe's Clinical Hematology, 12th edn., pp. 1082–1131. LippincoĴ Williams and Wilkins, Philadelphia (2009)

Demographic Features: Public Health in West Bengal – Current Status and Ongoing Interventions. gov.in/index_htm_files/Public%20 Health%20in%20 West%20Bengal.pdf

Dolai, T.K., Dutta, S., Bhattacharyya, M., Ghosh, M.K.: Prevalence of hemoglobinopathies in rural Bengal. India. Hemoglobin. **36**(1), 57–63 (2012)

Goh, L.P.W., Chong, E.T.J., Lee, P.C.: Prevalence of Alpha (α)-Thalassemia in Southeast Asia (2010–2020): A Meta-Analysis Involving 83,674 Subjects. Int. J. Environ. Res. Public Health **17**(20), 7354 (2020)

Jain, B.B., Roy, R.N., Ghosh, S., Ghosh, T., Banerjee, U., Bhattacharya, S.K.: Screening for thalassemia and other hemoglobinopathies in a tertiary care hospital of West Bengal: Implications for population screening. Indian J. Public Health **56**, 297–300 (2012)

Kamble, M., Chatruvedi, P.: Epidemiology of sickle cell disease in a rural hospital of central India. Indian Pediatr. **37**(4), 391–396 (2000)

Kumar, R., Gupta, S., Jindal, A., Kakkar, S., Kaur, A.: Screening of β-thalassemia trait and 295 other hemoglobinopathies among blood donors in Punjab. Int. J. Med. Public Health. **5**, 106–109 (2015)

Mandal, P.K., Maji, S.K., Dolai, T.K.: Present scenario of hemoglobinopathies in West Bengal, India: an analysis of a large population. Int. J. Med. Public Health. **4**, 496–499 (2014)

Modell, B., Darlison, M.: Global epidemiology of haemoglobin disorders and derived service indicators. Bull. World Health Organ. **86**(6), 480–487 (2008)

Mondal, S.K., Mandal, S.: Prevalence of thalassemia and hemoglobinopathy in eastern India: a 10- year high-performance liquid chromatography study of 119,336 cases. Asian J. Transfus Sci. **10**(1), 105–110 (2016)

Parthasarathy, V.: A search for Beta thalassemia trait in India. Turk. J. Haematol. **29**(4), 427–429 (2012)

Saxena, A., Phadke, S.R.: Thalassaemia control by carrier screening: the Indian scenario. Curr. Sci.. Sci. **83**, 291–295 (2002)

Shrivastav, A., Patel, U., Joshi, J.R., Kaur, A., Agnihotri, A.S.: Study of hemoglobinopathies and Hb variants in population of western India using HPLC: a report of 7,000 cases. J. Appl. Hematol. **4**, 104–109 (2014)

Tyagi, S., Saxena, R., Choudhry, V.P.: HPLC — how necessary is it for haemoglobinopathy diagnosis in India? Indian J. Pathol. Microbiol. **46**, 390–393 (2003)

Varawalla, N.Y., Old, J.M., Sarkar, R., Venkatesan, R., Weatherall, D.J.: The spectrum of beta- thalassaemia mutations on the Indian subcontinent: the basis for prenatal diagnosis. Br. J. Haematol. **78**(2), 242–247 (1991)

Verma, I.C., Choudhry, V.P., Jain, P.K.: Prevention of thalassemia: a necessity in India. Indian J. Pediatr. **59**(6), 649–654 (1992)

Weatherall, D.J.: Hemoglobinopathies 262 worldwide: present and future. Curr. Mol. Med. **8**(7), 592–599 (2008)

Weatherall, D.J.: The challenge of haemoglobinopathies in resource-poor countries. Br. J. Haematol. **154**(6), 736–744 (2011). https://doi.org/10.1111/j.1365-2141.2011.08742.x

Weatherall, D.J., Clegg, J.B.: Inherited haemoglobin disorders: an increasing global health problem. Bull. World Health Organ. **79**(8), 704–712 (2001)