Original Article

A need of awareness programs to reduce the incidence of beta thalassemia and other hemoglobinopathies in Navi Mumbai

Rao Rajiv[1*], Vidhate Deepali[2], Sukriti[3], Loungani Lavina[4]

{[1]Associate Professor, [3,4]Jr.Resident, Department of Pathology} {[2]Associate Professor, Department of Biochemistry}
Dr. D Y Patil University School of Medicine, Nerul, Navi Mumbai, Maharashtra, INDIA.
Email: rajiv.rao@dypatil.edu

Abstract

Haemoglobinopathies are most common inherited disorders of haemoglobin synthesis. Some Communities in India are vulnerable to many erythrocytic hereditary and haematological disorders. The aim of this study is to increase the awareness of Haemoglobinopathies in Navi Mumbai and to find the incidence of Beta Thalassemia. Results: The present study included 527 subjects of Indian descent with mean age of 27.50 (± 11.44) years and reported normal hemoglobin pattern in 469 cases (88.9%), Hemoglobinopathies in 44 cases (8.4%) and borderline A2 levels has been found in14 cases (2.7%).The prevalence of increase in Hb A2 levels above normal were highest i.e. 34 cases (6.4%) out of 527. In 4 cases (0.7%) HbS levels were higher than normal. Further we have reported 2 cases (0.4%) of increased levels of HbF and one case (0.2%) with increased HbE level. Increase levels of at least two hemoglobin fractions were reported in 4 cases. The prevalence of Beta Thalassemia trait was highest among the study subjects and amongst which two patients were first degree relatives. Further reported was 1 case (0.3%) of HbE Beta plus Thalassemia and 1 case (0.3%) of Sickle Beta plus thalessemia. Out of 527cases 322 subjects underwent investigations for hematological parameters and 32 (9.9%) cases showed abnormal hemoglobin fractions. A significant correlation was noted between Beta thalessemia trait patients and microcytic hypochromic anemia. Conclusion: A high prevalence of Beta thalessemia trait with low incidence of other haemoglobin variants was observed in the study population. Awareness about the hemoglobinopathies and its screening must be implemented as a preventative measure.

Keywords: Haemoglobinopathies, Beta thalassemia, microcytic hypochromic anemia.

[*]**Address for Correspondence:**
Dr. Rao Rajiv, Associate Professor, Department of Pathology, Dr. D Y Patil University School of Medicine, Nerul, Navi Mumbai, Maharashtra, INDIA.
Email: rajiv.rao@dypatil.edu
Received Date: 01/12/2016 Revised Date: 01/12/2016 Accepted Date: 01/12/2016

Access this article online	
Quick Response Code:	Website: www.statperson.com
	DOI: 04 December 2016

INTRODUCTION

Prevalence of Haemoglobinopathies is increasing and 7% of the world's population being carriers of Hemoglobinopathies [WHO report 2006][1]. Early detection and characterization of the haemoglobinopathies plays an important role in changing the prevalence pattern. Appropriate counseling to couples and families who may be at risk of severe haematological consequences became an essential part of this. A broaden awareness program with knowledge of basic diagnosis, counselling and management of the haemoglobin disorders among population is essential to deal with Hemoglobinopathies[2,3]. Identification of carriers and frequencies of these inherited characters have been reported in various populations and ethnic groups in India[4-8]. In several regions of India haemoglobinopathies are found during a premarital screening. Beta Thalassemia has been reported as the major syndrome with high percentage of prevalence in Gujarat, Assam, Orissa, Maharashtra, and Kerala due to residing local tribal population[4-7]. Thalassemia is an autosomal recessive inherited group of disorders of hemoglobin

How to site this article: Rao Rajiv, Vidhate Deepali, Sukriti, Loungani Lavina. A need of awareness programs to reduce the incidence of beta thalassemia and other hemoglobinopathies in Navi Mumbai. *International Journal of Recent Trends in Science and Technology.* December 2016; 21(2): 110-113. http://www.statperson.com (accessed 07 December 2016).

synthesis characterised by the absence or reduction of one or more of the globin chains of hemoglobin. Children suffering with Beta Thalassemia major show severe anemia and needs regular transfusion for survival from early childhood[8]. In 1910 characteristic sickle shaped red cells were described by a physician and Linus Pauling and his colleagues showed its altered electrophoretic mobility pattern[9,10]. A single base A>T mutation in the sixth residue of the β-globin chain, leads to a substitution of valine for glutamic acid and results in abnormal haemoglobin S (HbS).In India a high prevalence of sickle gene among tribal populations has been observed in different States[11]. After birth, HbF synthesis rapidly declines and HbF is gradually substituted by Hb A in the peripheral blood. Hence in first two years of life, very low levels of HbF (less than 1%) were found. HbE was the fourth abnormal haemoglobin to be identified by haemoglobin electrophoresis, in 1954[12], and in 1961 it was characterised as having the substitution of lysine for glutamic acid at position 26 of the β-globin chain[13]. Many types of HbE syndromes have been observed due to various interactions with α- thalassaemia, β-thalassaemia or other haemoglobin variants. Geographically, it is prevalent in South-East Asia and the north-eastern states of India. HbE variants usually manifest as homozygous HbE disease, heterozygous HbE (HbE trait), and doubly heterozygous forms HbE-β thalassaemia[14]. Hence it is the need of the hour to identify the carriers of these disorders and to take preventive measures by prenatal diagnosis and counselling to couples at risk of having a child with haemoglobinopathies.

MATERIAL AND METHODS

The study was conducted at D. Y. Patil University School of Medicine and D. Y. Patil Hospital and Research Center, Nerul, Navi Mumbai, India over a period of 2 years. Subjects of age group between 04 Months to 55 years were randomly selected from Out-patient-department (OPD) and Inpatient department (IPD) on the basis of inclusion and exclusion criteria (including the suspected cases of hemolytic anemia without any systemic /chronic infection and drug abuse). An informed consent was obtained from the participants along with the questionnaires containing information related with age, sex, resident, nationality, educational qualification, occupation and financial status etc. Blood samples were collected in ethylene diamine tetrachloride acetate (EDTA) vials and run on automated cell counter for complete blood counts as indicated. For each patient, a peripheral blood smear (PBS) was prepared and stained with Leishmann stain and observed microscopically for red cell morphology for the supporting of diagnosis of hemoglobinopathies. Hb variant analysis was studied for various hemoglobinopathies and variants by BioRad D-10, an automatd cation exchange HPLC instrument that has been used to quantify Hb A2, Hb F, Hb A along with screening Hb variants like Hb S Hb D and Hb E.

RESULTS

The present study included 527 subjects of Indian descent with mean age of 27.50(± 11.44) years, of which 469 were normal (88.9%), 44 cases of Hemoglobinopathies (8.4%) and 14 cases were found to be on borderline A2 levels (2.7%).The prevalence of increase in Hb A2 levels above normal were highest in 34 cases (6.4%) out of 527, while 14 cases (2.7%) of HbA2 were found to be borderline. In 4 cases (0.7%) HbS levels were higher than normal. We have reported 2 cases (0.4%) of increased levels of HbF and one case (0.2%) with increased HbE level. Further, 4 cases were also reported with increased levels of at least two hemoglobin fractions. The prevalence of Beta thalassemia trait was highest among the study subjects and amongst which two patients were first degree relatives. Further reported was 1 case (0.3%) of HbE Beta plus Thalassemia and 1 case (0.3%) of Sickle Beta plus thalassemia.

Table 1: Distribution of hemoglobin patterns in the study population

Variables	Mean	S.D.(±)
Age	27.50	11.44
Hb A	95.55	10.44
HbA2	2.76	1.06
Hb F	0.83	7.09
Hb S	0.43	4.51
Hb E	0.05	1.07

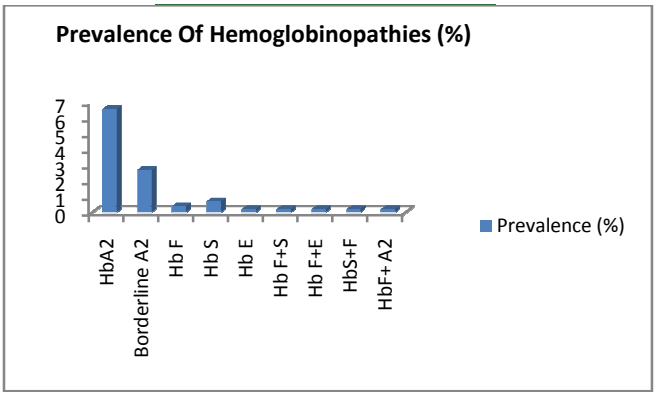

Figure 1: Prevalence of Haemoglobinopathies

Table 2: Comparison of hemoglobin patterns in the study population

Variables	Control (n=345)		Hemoglobinopathies (n=34)		Significance
	Mean	S.D. (±)	Mean	S.D. (±)	
Hb A	97.01	4.7	81.69	26.5	0.000
Hb A2	2.54	0.54	4.75	2.40	0.000
Hb F	0.14	1.93	8.40	22.59	0.000
Hb S	0.07	1.58	4.38	14.29	0.000
Hb E	0.00	0.00	0.69	3.69	0.000

Table 2: Comparison of CBC parameters andhemoglobin patterns

Variables	Control (n=290)		Hemoglobinopathies (n=32)		Significance
	Mean	S.D. (±)	Mean	S.D. (±)	
Hb (g/dl)	9.4	2.5	8.1	1.9	0.43 (NS)
RBC (10˙6/µl)	4.0	0.81	4.0	1.1	>0.05(NS)
PCV (%)	30.89	7.4	26.45	6.8	0.07(NS)
MCV (fL)	77.80	13.88	64.7	13.22	0.04
MCH (pg)	24.48	5.69	21.51	4.82	0.02
MCHC	31.00	2.38	31.26	1.30	0.21
Hb A	97.01	6.03	77.87	29.75	0.02
HbA2	2.29	0.43	4.5	2.7	0.00
Hb F	0.21	2.4	11.5	25.89	0.00
Hb S	0.1	2.0	6.03	16.52	0.00
Hb E	0.0	0.0	0.95	4.32	0.00

*Statistically significant p<0.005; NS- Statistically Non Significant

DISCUSSION

The present study reported 45 cases (8.5%) of Hemoglobinopathies out of a total of 527 from local population. 34 cases (6.4%) were observed to have Beta Thalassemia trait. Sachdev *et al*[15] in his study on 2600 cases for identification of various hemoglobin disorders observed 327 cases with abnormal hemoglobin fractions out of which 232 cases (8.9%) were diagnosed to have Beta Thalassemia Trait. In our study we also reported 14 patients (2.7%) with borderline HbA2 level. In 2007, Colah *et al* (16) observed 7 cases of borderline Hb A2 levels and further reported that amongst them 5 cases were silent carriers. Out of 527 cases, 322 cases were also screened for anemia and majority with abnormal Hemoglobin fractions were found to have Microcytic Hypochromic Anemia. Similarly Bhalodia *et al*[17] in his study on 500 cases, observed 43 cases with abnormal hemoglobin fraction and majority having Microcytic Hypochromic anemia. Wajchman *et al*[18] in his study stated that the key element in the diagnosis of Beta Thalassemia Trait is the presence of non-iron deficient Microcytic Hypochromic Anemia. In our study we observed 4 cases with sickle cell trait. Recently Colah *et al* (2015) and Kaur *et al* (2013) carried out an extensive study for assessing the incidence of sickle cell anemia in tribal population of India and reported a high incidence. Further it was suggested that it may be due to intermittent marriage system in these tribs[19,20]. Madan *et al*[21] in the study of Beta Thalassemia and hemoglobinopathies in India observed that HbS trait was seen in children in Mumbai, probably due to greater influx of population from tribal areas. The wide prevalence of thalassemias and hemoglobinopathies has been attributed to migration of people from one region to another and consanginous marriages in certain sub populations like Parsis and Muslims in our population. With increasing awareness, detection of these disorders in some countries mostly occurs during premarital screening[22-25]. In western European countries detection usually occurs through preconceptional and neonatal screening programs[27]. Conclusion: A high prevalence of Beta thalessemia trait with low incidence of other hemoglobin variants was observed in the study population. Awareness about the hemoglobinopathies and its screening must be implemented as a preventative measure especially in the child baring age group. We strongly recommend the inclusion of screening for Hemoglobinopathies as a compulsory first trimester investigation in all primigravidae.

REFERENCES

1. Prevalence of Haemoglobinopathies. Who- Executive Board EB118/5, 118th Session Report by the Secretariat on Thalassaemia and other haemoglobinopathies. 2006; 1-8.
2. Jain BB, Roy RN, Ghosh S, Ghosh T, Banerjee U and Bhattacharya SK. Screening for thalasemia and other hemoglobinopathies in a tertiary care hospital of West Bengal: implications for population screening. Indian Journal of Public Health 2012; 56(4): 297-300.
3. Chopra GS, Nair V, Gupta PK, Mishra DK, Sharma A and Mathew OP. Spectrum of haemoglobinopathies in a tertiary care hospital of armed forces. Medical Journal Armed Forces India. 2008; 64(4): 311-314.
4. Patel J, Patel A, Patel J, Kaur A and Patel V. Prevalence of Haemoglobinopathies in Gujarat, India: A Cross-Sectional Study. The Internet J Hematol. 2009; 5: 1.
5. Patel J, Shrivastav A, Joshi JR, Agnihotri AS, Kaur A and Thakkar B. Detection of hemoglobinopathies and thalasemias in population of Gujarat State using HPLC: Analysis of 2022 cases. Pathology and Laboratory Medicine 2012; 4(2): 80-84.
6. Balgir RS. Spectrum of haemoglobinopathies in the state of Orissa, India: A ten years cohort study. J Assoc Physicians India 2005; 53: 1021-1026.
7. Jain BB, Roy RN, Ghosh S, Ghosh T, Banerjee U and Bhattacharya SK. Screening for thalasemia and other hemoglobinopathies in a tertiary care hospital of West Bengal: implications for population screening. Indian Journal of Public Health 2012; 56(4): 297-300.
8. Balgir RS. The burden of haemoglobinopathies in India and the challenges ahead. Curr Sci 2000; 79:1536-1547.

9. Stuart MJ, Nagel RL. Sickle cell disease. Lancet. 2004; 364:1343–60.
10. Rees DC, Williams TM, Gladwin MT. Sickle cell disease. Lancet. 2010; 376: 2018–31.
11. Rao VR. Genetics and epidemiology of sickle cell anemia in India. Indian J Med Sci. 1988; 42: 218–22.
12. Nagel R, Steinberg MH. Haemoglobin SC disease and HbC disorders In: Steinberg MH, Forget BG, Higgs DR, Nagel RL, editors. ed. Disorders of Haemoglobin: Genetics, Pathophysiology, and clinical management Cambridge: University Press; 2001. 756 785.
13. Itano HA, Bergren WR, Sturgeon P. Identification of fourth abnormal human haemoglobin. Journal of the American Chemistry Society. 1954; 76: 2278–2282.
14. Aggarwal S, Saluja S, Bhasin S, Sharma M, Gupta DK, Gupta B, Mittal V. HbE variants–retrospective analysis in a tertiary care centre. Journal, Indian Academy of Clinical Medicine. 2011 Oct; 12(4).
15. Sachdev R, Dam AR, Tyagi G. Detection of Hb variants and hemoglobinopathies in Indian population using HPLC: report of 2600 cases. Indian journal of pathology and microbiology. 2010 Jan 1; 53(1):57.
16. Colah R B, Surve P, P. Sawant, E. D'Souza, K. Italia, S. Phanasgaonkar, A.H. Nadkarni and A.C. Gorakshakar. HPLC Studies in Hemoglobinopathies. Indian Journal of Pediatrics, Volume 74—July, 2007.
17. Bhalodia JN, Oza HV, Modi PJ, Shah AM, Patel KA, Patel HB. Study of Hemoglobinopathies in Patients of Anemia using High Performance Liquid Chromatography (HPLC) in Western India. Community Med. 2015;6(1):35-40.
18. Wajcman H, Moradkhani K. Abnormal haemoglobins: detection and characterization. The Indian journal of medical research. 2011 Oct 1; 134 (4):538.
19. Colah R, Mukherjee M, Martin S and Ghosh K. Sickle cell disease in tribal populations in India. Indian J Med Res 141, May 2015, pp 509-515.
20. Kaur M, Dangi CBS, Singh M, Singh H, Kapoor H. Burden of sickle cell disease among tribes of India : A burning problem. Int Res J Pharm App Sci 2013; 3: 60-80.
21. Madan N, Sharma S, Sood SK, Colah R, Bhatia HM. Frequency of β-thalassemia trait and other hemoglobinopathies in northern and western India. Indian journal of human genetics. 2010 Jan 1; 16 (1):16.
22. Colah RB, Gorakshakar AC. Thalassemias : Molecular genetics in antenatal diagnosis. In Lokeshwar MR, Marwaha RK, Shah M. eds. Recent advances in Pediatrics. New Delhi; Jaypee Brothers, 2000: 22-45.
23. Karimi M, Jamalian N, Yarmohammadi H, et al. Premarital screening for β-thalassaemia in Southern Iran: options for improving the programme. J Med Screen. 2007; 14: 62–66.
24. Cowan RS. Moving up the slippery slope: mandated genetic screening on Cyprus. Am J Med Genet C Semin Med Genet. 2009; 151C:95–103.
25. Keskin A, Turk T, Polat A, et al. Premarital screening of beta-thalassaemia trait in the province of Denizli, Turkey. Acta Haematol. 2000; 104: 31–33.
26. Colah RB, Gorakshakar AC. Thalassemias : Molecular genetics in antenatal diagnosis. In Lokeshwar MR, Marwaha RK, Shah M. eds. Recent advances in Pediatrics. New Delhi; Jaypee Brothers, 2000: 22-45.
27. Benson, J. M.; Therrell, B. L. (2010). "History and Current Status of Newborn Screening for Hemoglobinopathies". Seminars in Perinatology. 34 (2): 134–144.

Source of Support: None Declared
Conflict of Interest: None Declared