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STUDY OF PREVALENCE OF HEMOGOBINOPATHY CONDITIONS IN PATIENT POPULATION OF TERTIARY CARE HOSPITAL AT NAVI-MUMBAI

Biochemistry

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ABSTRACT

Background: Prevalence of different hemoglobinopathy condition varies state wise in India due to ethnic diversity. In state of Maharashtra especially in Riagad district, the data on hemoglobinpothy is scarce. This district is mainly populated by Agri and some tribal communities. **Methodology:** A pilot study was undertaken to determine the prevalence of hemoglobinopathies in patients visiting our laboratory of tertiary care hospital located in Panvel taluka of Raigad district. Total 1675 blood samples received from January 2015 to May2017 at the MGM Medical College and Hospital's Central Laboratory were screened.

Result: We diagnosed total 166 (9.91%) patient with different hemoglobinapathy conditions from the screened samples. we reported maximum patients suffering β thalassemia minor (4.48 %), where as other conditions like β thal major, Sickle cell disease prevalence varied from 0.5 % To 2. Thus mass screening, Pre marital counselling, awareness programs, of the population especially childbearing age and school going children will help in early detection various hemoglobinopathy conditions.

KEYWORDS

Hemoglobinopathies, β thalassemia, sickle cell anemia, Prevalence

Introduction

Haemoglobinopathies are the commonest hereditary disorders in India and pose a major health problem. The data on the prevalence of β-thalassemias and other haemoglobinopathies in different caste/ethnic groups of India is scarce.

β- thalassemias and their interaction with haemoglobin E(HbE) and haemoglobin S (HbS) are a considerable health problem in India and contribute significantly to morbidity and mortality. Earlier studies have shown that the overall prevalence of β-thalassemia is 3–4 %. [1,2]

The overall prevalence of β -thalassemia in India is 3-4% with an estimate that around 10,000-12,000 children are born every year with β -thalassemia major. A recent study in India showed that the overall prevalence of β -thalassemia trait[3] was 2.78 % and varied from 1.48% to 3.64% in different states whereas prevalence of sickle cell anemia reported was 1.44%. Data on prevalence of other hemoglobinopathies is scarce.[4]

The most common techniques used for diagnosis of hemoglobinopathies in India includes clinical and family history, red cell indices, complete blood counts, Hemoglobin (Hb) F estimation, sickling test and Hb electrophoresis. These methods are conventional and has their own limitations for eg identification of Hb variants with same electrophoretic mobility by Hb electrophoresis and also diagnosing certain compound heterozygous states[5]. These problems can be overcome by, cation exchange high-performance liquid chromatography (CE-HPLC) by Biorad D 10 of variant which estimates various normal and abnormal Hb fractions[6]. It is considered as a gold standard method for accurate detection of hemoglobinopathies, thereby aiding in better diagnosis of hemoglobinopathies and management.[7]

This study was undertaken to find prevalence of different hemoglobinopathy conditions in patients referred to Central Clinical Laboratory of MGM Hospital located in Panvel taluka of Raigad district.

Materials and Methods

This study included analysis of 1675 blood samples received from January 2015 to May 2017 at the MGM Medical College and Hospital's Central Laboratory, which caters IPD and OPD patients of the MGM Hospital. Patients visiting medicine OPD with anemia, generalized weakness, fever, splenomegaly, who were suspected hemoglobinopathy on the basis of clinical and family history were referred to laboratory for investigations. The investigation advised by the clinicians mainly were Complete blood count with peripheral smear and Hemoglobinopathy screening. Patients with a history of recent blood transfusion and inadequate sample were excluded from the study. Three milliliters of intravenous blood was collected in dipotassium ethylenediamine tetraacetic acid vacutainers and was run in XN 1000 for hemogram and red cell indices; the same was used for HPLC. The samples were run on instrument D10 manufactured by Bio-Rad Laboratories. The instrument works on the principle of HPLC. An HbA_2 and HbF calibrator and two levels of controls were analyzed at the beginning of each run. Based on retention time and proportion of Hb variants,different hemoglobinopathies were diagnosed and their prevalence was analyzed. HbA2 level of > 3.5 % was used as a cutoff fordiagnosis of β-thalassemia carriers. RBC indices were measured on a haematologycounter (XN 1000, 6 part differentail).

Results

Laboratory records of total 1675 patients analysed for suspected hemoglobinopathies were screened. Total 166 (9.91%) patients were found to be sufferers of different hemoglobinapathies condition, out of which 79 were males and 87 were females. The age group ranged from 01 to 50 years with a maximum number of patients in the age group 1-20 years. Screening tests for in these cases included CBC, Sickling tests and Meisner's index. It was observed that in patients with hemoglobinopathy conditions showed MCV < 83 fl and MCH < 27 pg.

All patients were between age group of 1-40yrs. We reported patients with beta thalassemia major and minor, patients suffering from sickle cell disease with beta thalassemia and also with Sickle cell homozygous disease. We also reported few cases with Hb E trait and Hb E homozygous sickle cell disease homozygous.

Out of total 166 Hemoglobinopathies condition reported, 28 (1.67 %)patients were with β thalassemia major,75 (4.48 %) patients with β thalassemia minor, 29 (1.73 %) patients with Sickle cell diseases with thalassemia trait, 16 (0.96 %) patients with sickle cell disease, 12 (0.72 %) with Hb E thalassemia and 6 (0.35 %)with Hb E homozygous.

Table 1 Showing age wise spectrum of hemoglobinopathies

Subjects with hemoglobinopathy Variable	Variable	No. of subjects	Prevalence
β thalassemia Trait and delta β thalassemia	>20 yrs	49	2.92
	21-35 yrs	21	1.25
	36-50 yrs	05	0.29
β thalassemia major	>20 yrs	24	1.47
	21-35 yrs	04	024
	36-50 yrs	-	-
Sickle Cell Disease with β thalassemia	>20 yrs	19	1.13
	21-35 yrs	10	0.6

	36-50 yrs	-	-
Sickle Cell Disease homozygous	>20 yrs	11	0.66
	21-35 yrs	05	0.30
	36-50 yrs	-	-
HB E β thalassemia	>20 yrs	06	0.36
	21-35 yrs	06	0.36
	36-50 yrs	-	-
HB E homozygous	>20 yrs	03	0.18
	21-35 yrs	02	0.12
	36-50 yrs	01	0.06

Table 2 Gender wise distribution of hemoglobinopathies in study subjects

Hemoglobinopathy condition	Males	Females
β thalassemia Trait and delta β thalassemia	16	12
β thalassemia major	36	39
Sickle Cell Disease with β thalassemia trait	12	17
Sickle Cell Disease homozygous	11	05
HB E β thalassemia	10	02
HB E homozygous	06	-

Table 3 Showing Percentage of Hemglobinpathies in study subjects

Name	Total no of cases	Percentage
Non-hemoglobinopathies	1509	90.08
Hemoglobinopathy Conditions		
β Thalassemia major	28	1.67
β Thalassemia minor	75	4.48
Sickle Cell Disease with β thalassemia trait	29	1.73
Sickle Cell Disease Homozygous	16	0.96
HB E thalassemia	12	0.77
HB E homozygous	06	0.35
Total Subjects	166	9.91

Table 4 Comparison of hematological indices in different hemoglobinopathy conditions

Hemat-ological indices	Normal Mean ±SD	β Thalassemia minor	β Thalassemia major	Sickle Cell Disease with β thalassemia	Sickle Cell Disease Homozygous	HB E thalassemia	HB E homozygous
Hb gm%	10.60 (2.41)	8.25 ± 3.1	5.21 ± 3.9	8.01± 2.4	6.69±1.09	4.0 ±3.9	7.9 ±4.2
RBC	4.00 (0.74)	4.3 ±1.2	2.17 ± 0.67	4.41±1.3	43.39±	2.46 ± 0.31	4.37 ± 0.49
MCV (fL)	76.80 (11.20)	64.0 ±6.1	61.33 ± 5.9	60.05±5.6	59.7 ± 7.1	58.6 ± 4.6	50 ± 3.8
MCH (pg)	26.70 (4.72)	24 ± 2.89	22.32 ± 2.7	19.37± 3.0	18.67± 2.91	18.9 ±0.9	20.3 ± 1.76
MCHC (g/dL)	34.40 (2.69)	33.9 ± 3.4	36.7 ± 3.4	30.8 ± 2.6	29.12 ± 4.13	30.8 ±2.6	36.9 ± 3.01

Fig 1 Chromatogram of Hb E Trait

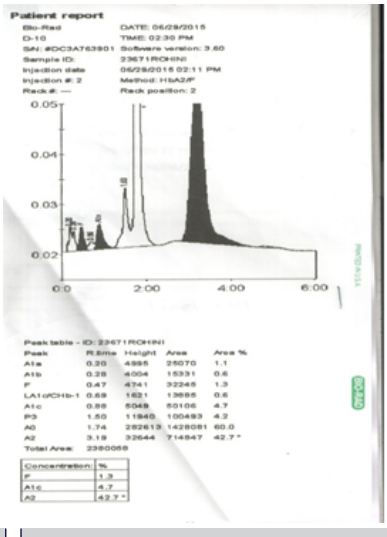


Fig 2 Chromatogram of Delta β thal Trait

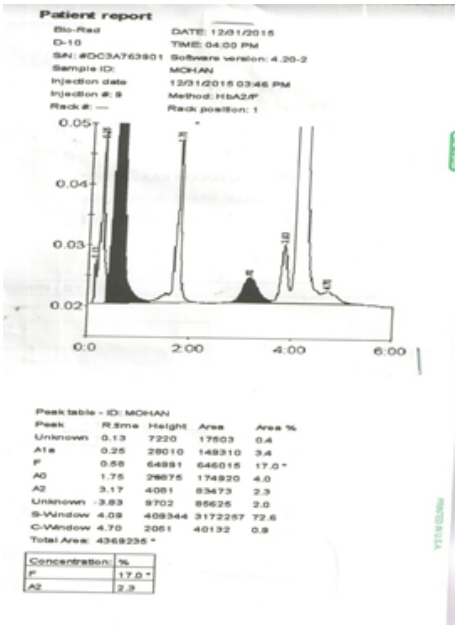


Fig 3 Chromatogram of β- Thal major

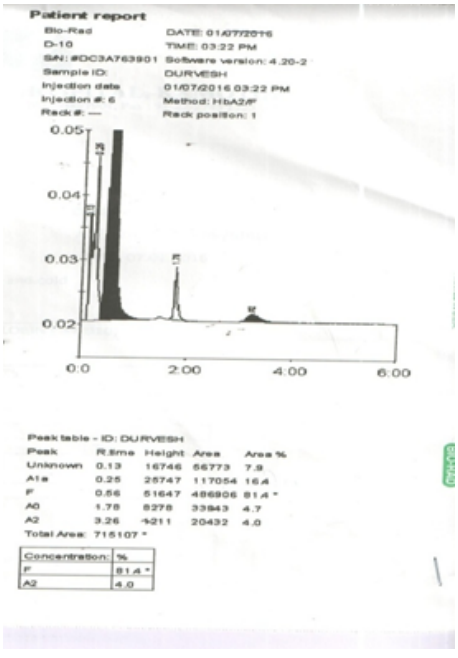


Fig 4 Chromatogram of Hb S homozygous

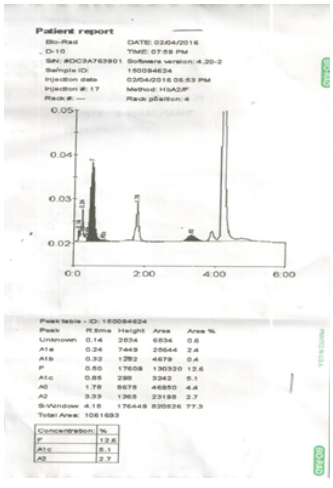


Fig 5 Chromatogram of HbE homozygous

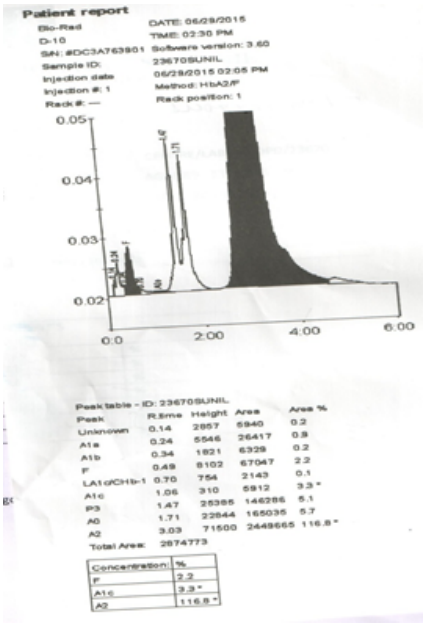
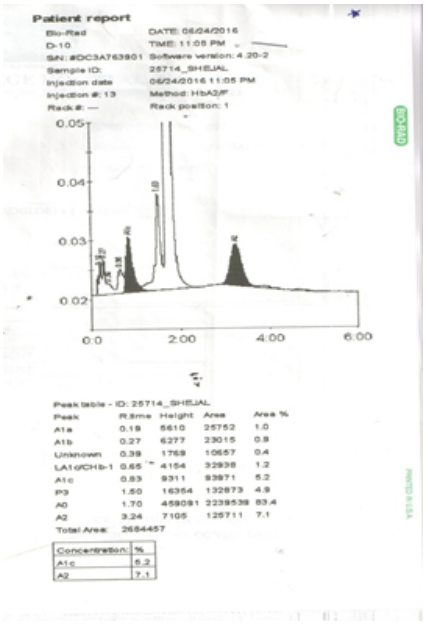


Fig 5 Chromatogram of β Thal Minor



Discussion

In our study, all screened 1675 patients were from Panvel Taluka in Raigad district,out of which 166 (9.91%) were diagnosed of different hemoglobinopathy conditions. Our study also showed good correlation between hematological indices and A2/ F percentage.

We compared prevalence of hemoglobinopathy conditions obtained b us with finding of reported other authors and observed that findings of our study are in accordance with Verma et al who revealed higher prevalence of hemoglobinopathies in males 110/804 (13.7%) as compared to females 33/376(8.8%)[8]. Finding of our study are very much consistent with Sachdeva et al, They reported 8.9 % cases of the beta thalassemia trait, 0.6% cases of beta thalassemia major. The 0.5% cases of Hb D Punjab, and seven cases of Hb E which included two Hb E homozygous and five Hb E heterozygous.[9]

Our findings are contradictory with the prevalence of hemoglobinopathies in Gujarat, reported by Patel et al., They have mentioned in their study that out of 428 subjects, 153(35.7%) had hemoglobinopathies.[10] while another study conducted by them in year 2011 stated higher prevalence of 38.97%.[11]. Ambekar et al. reported the frequency of hemoglobinopathies in Western Maharashtra of 26.5.[12] Chopra et al. revealed that out of 1032 participant, 258 (25%) cases had abnormal hemoglobin.[13] Also study done by Panda A et al. based on West Bengal population illustrated that prevalence of hemoglobinopathies was 20.47%.[14] Rao et al,2010 observed 32.4% males and 67.6% females were suffering from hemoglobin disorders[15] while Uddin et al, 2012 reported an equal incidence of hemoglobinopathies in both males and females.[16]

Thus a closer look at hemoglobin indices and a detailed peripheral blood picture study may help in timely detection of different hemoglobinopathy conditions.

It is also important in preventing birth of homozygous thalassemia major child by genetic counsel-ling regarding the nature of the disease. A thalassemia major child is dependent of regular blood transfusions to maintain life since early childhood. However safe blood is available only for a small fraction and most such patients die due to iron overload.

Normally hemoglobinopthy conditions get presented in the form of mild to severe anemia, hence detection of these condition demands careful evaluation of hemoglobin indices and clinical examination of patient. We diagnosed 29 cases (1.67%) of sickle cell trait and 16 cases of (0.96%) sickle cell disease during our study in anaemic patients. Timely detection of sickle cell trait can be helpful in warning patients about possible complications and to plan preventive measures. Prenatal diagnosis of sickle cell disease helps in planning treatment and preventive modalities.

Thus identification of Hb variants at the earliest is of utmost importance for hemoglobinopathy conditions which may include Heterozygous or homozygous state of HbE and Beta thalassemiais. This can be achieved by increasing awareness of this rare disorder, in clinicians and patients to assist in prenatal diagnosis, genetic and clinical man-agement. Premarital counseling and screening is also an essential element in eradication of hemoglobinopathies but it is still considered taboo. So the best approach would be to target those patients attending the hematology OPD, the antenatal population and extended family members. Person having positive report for carrier state should be counseled regarding the nature of the disease and implications of being carrier which may help in preventing birth of child with homozygous inheritance of hemoglobinopathies.

A multipronged approach including screening of high school/college students, premarital screening, and of the extended family of thalassemics along with antenatal diagnosis needs to be considered for this vast and ethnically diverse country. Education and awareness regarding thalassemics need to be accelerated among medical practitioners, paramedics, the thalassemic and general population to reduce the morbidity and mortality. This may help in reducing financial and sociopsychological burden of the thalassemic families.

Conclusion

Our study was an pilot study which mainly screened patients visiting our hospital and determined prevalence of hemoglobinopathies in patient population located in Panvel taluka of Raigad district. The data regarding prevalence and distribution can be useful in planning preventive measure ans awareness strategy for management of various hemoglobinopathies conditions in patients visiting MGM Hospital OPD.

Limitations:-

The study population included screening of data of patients visiting MGM central Laboratory. The determination prevalence of different hemoglobinopathy condition should be sought from much larger sample size.

Acknowledgement

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Competing Interests

The authors have no competing interests.

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