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THE STUDY OF PREVALENCE OF HEMOGLOBINOPATHIES IN ANTENATAL PATIENTS AT TERTIARY CARE HOSPITAL.

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Abstract:
Introduction: Hemoglobinopathies affects aminoacid sequence of globin chain biosynthesis either quantitatively or qualitatively. These disorders are the most common mendelian genetic diseases and manifest as hemolytic anemias. Hence identifying hemoglobinopathies during the antenatal period is essential to assess the vertical transmission in foetus. **Aim :**This study is designed to find out the prevalence of various hemoglobinopathies in antenatal women by High performance liquid chromatography(HPLC) and its correlation with RBC indices. **Material &Method :** We conducted a observational study for one year from January 2023 to December 2023. The study included all the antenatal women whose blood samples were received in the hematology department for HPLC. The cases with abnormal HPLC findigs were analyzed for hematological parameters including hemoglobin, RBC count and RBC Indices[mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH), mean corpuscular hemoglobin concentration (MCHC) & red cell distribution width-coefficient of variation (RDW-CV)]. **Result :**During the study period, total 716 antenatal cases were analyzed for HPLC and RBC indicies. Hemoglobinopathies were detected in 58 antenatal cases(8.3%). Out of total 58 cases of hemoglobinopathies, 42 cases of β-thalassemia trait (72.4%), 8 cases of Hb D Punjab trait(13.8%), 5cases of Hb S trait (8.6%), 02 cases of Hb E trait (3.5%), 01 case of Hb D Iran trait(1.7%). **Conclusion:** Red blood cell (RBC) indices, MCV and MCH can be used for initial screening of hemoglobinopathies followed by more confirmatory methods (HPLC). Detecting carrier stage of hemoglobinopathies is crucial for decision making in antenatal women and assessment of hemoglobinopathies transmission in offsprings.
Keywords: Antenatal, Hemoglobinopathies, HPLC, β-thalassemia trait.

INTRODUCTION:

Thalassemia and hemoglobinopathies are a group of inherited conditions characterized by abnormalities in the synthesis or structure of hemoglobin (Hb). According to various estimates, approximately 7% of the world population is a carrier of Hb disorders, leading to high morbidity and mortality. Therefore, it is pertinent to detect the carrier stages as early as possible to prevent disease progression and halt the inheritance to further generations. The clinical presentation of hemoglobinopathies varies from being completely asymptomatic to severe transfusion-dependent anemia. Investigations to determine the genetic burden of Hb disorders during antenatal care are thus the need of the hour. The routine hematological parameters including RBC indices provide significant clues which can help in early diagnosis. Other advanced techniques like high-performance liquid chromatography (HPLC) should also be done to diagnose thalassemia and hemoglobinopathies, especially in antenatal/females of reproductive age group screening[1,2,3].

Materials and methods:

This was a observational study carried out for 1 year(from January 2023 till December2023).All the antenatal women whose blood samples were received in the hematology laboratory for HPLC were included. Screening of hospital computer database was done to retrieve records about relevant clinical information and investigations.

HPLC was performed using the Biorad variant D-10 instrument. HPLC report and chromatogram were generated for every case where different peaks were identified in defined windows with relevant information like retention time, the relative percentage of hemoglobin fractions, and total area.

An elaborative hematological workup including hemoglobin, RBC count, and RBC indices [(Mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH), mean corpuscular hemoglobin concentration (MCHC), & red cell distribution width-coefficient of variation (RDW-CV)] was carried out in all the cases with abnormal HPLC findings. It was performed on a 7-part differential automated hematology analyzer (Siemens advia 2120i) which works on the principle of electrical impedance and light scattering.

Results:

During the study period, a total of 716 antenatal females (mean age 29.68 ± 10.9 years) were screened. Of the 716 females, 58 displayed (8.1%) abnormal HPLC patterns concerning Hb A2, Fetal hemoglobin (HbF), or abnormal peaks.
Of the 58antenatal women, the majority (n = 42; 72.4%%) were affected with β-thalassemia trait, followed by HbD Punjab trait (n = 8; 13.8%), HbS trait (n = 5; 8.6%), HbE trait (n = 2; 3.5%) and HbD Iran trait (n = 1; 1.7%) as mentioned in Table 1.

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[Table-1]: Spectrum of thalassemia and other hemoglobinopathies.

Sr. no.	Type of hemoglobinopathy	No. of patients out 0f 58 cases	Percentage (out of abnormal HPLC cases)	Percentage of hemoglobinopathy out of total antenatal cases 716(%)
1	Beta-thalassemia trait	42	72.4%	5.86
2	Hb -D Punjab trait	08	13.8%	1.12
3	Sickle cell trait	05	8.6%	0.70
4	Hb E trait	02	3.5%	0.28
5	Hb D Iran	01	1.7%	0.14

Table 2 shows the hematological data expressed as mean± standard deviation of common hemoglobinopathies found in this study. The mean hemoglobin values were low in women with β-thalassemia trait and sickle cell trait as compared to the HbD Punjab trait, whereas the MCV, MCH values were higher in HbD Punjab trait and Sickle cell trait when compared with β- thalassemia trait.

[Table-2]:Hematological parameters of common hemoglobinopathies in the study.

	Beta-thalassemia trait Mean± SD	Hb-D Punjab trait Mean± SD	Sickle cell trait Mean± SD
RBC count (million/µl)	4.75±0.74	4.61±0.67	4.12±0.59
Hb (gm/dl)	10.1±1.17	12.3±1.26	10.68±1.14
MCV(fl)	69.30±9.94	79.8±5.68	78.68±7.95
MCH (pg)	21.87± 4.22	26.9±1.80	26.12±3.26
MCHC (gm/dl)	31.4±1.71	33.68±1.58	33.16±1.45
RDW-CV (%)	17.36±2.54	17.0±4.76	16.72±3.17

Discussion:

Hemoglobinopathies are autosomal recessive inherited disorders. It includes two main groups, (1)thalassemias,with defects in the synthesis of a globin chain and (2) structural variants, with defects in the structure of hemoglobin. There is also an overlap between these groups called compound heterozygous state.[4]The birth of a β-thalassemia major or sickle cell anemia child poses an emotional and financial burden on both family and society. Whereas the compound heterozygous state also affect the quality of life of the child. This emphasizes the fact that detecting hemoglobinopathies during carrier state is necessary. The screening can be best done in the premarital stage, but it may not be possible in all the communities and ethnic groups, so the ideal candidate for this would be pregnant women.Routine hematological parameters like RBC indices along with HPLC can detect carrier state of different hemoglobinopathieswhich are clinically silent[5].The variations in the spectrum of different hemoglobinopathieswith regards to regions are known, but the data remains limited in this context.[3,6,7]

India bears a huge burden of hemoglobinopathies and the most prevalent is thalassemia.Yet, only a few studies are available providing data related to various hemoglobinopathies in terms of incidence, prevalence, morbidity, and mortality. Herein, we present the data in terms of the spectrum and prevalence of various hemoglobinopathies at our center. The spectrum of various hemoglobinopathies was in accordance with that of a study conducted by Narang V. et al. and Mohanty et al[6,8,9].

[Table-3]: Comparison of proportion of various hemoglobinopathies with different studies.

Type of Hemoglobinopathy	Proportion (%)-present study	Proportion (%)- Narang V et al (Punjab, north india)	Proportion (%)- bhukhanvala et al (Gujarat, western india)
Beta-thalassemia trait	42/58(72.4%)	122/169(72.1%)	102/174(58.63%)
Hb -D Punjab trait	08/58(13.8%)	30/169(17.8%)	11/174(6.32%)
Sickle cell trait	05/58(8.6%)	2/169(1.2%)	46/174(26.44%)
Hb E trait	02/58(3.5%)	1/169(0.6%)	07/174(4.02%)
Other	01/58 (1.7%)	14/169(8.3%)	08/174(4.59%)

Table 3 is showing proportion(%) of various hemoglobinopathies out of total cases of hemoglobinopathies and their comparison with similar studies performed earlier. Our study has revealed that the proportion of β-thalassemia trait is higher 72.4% and findings were comparable to studies by Narang V et al (Punjab, North India) and bhukhanvala et al[8,9]. However, the prevalence of the total hemoglobinopathies (8.10%) and β-thalassemia trait (5.86%) was higher in our study as compared to Narang V et al. total hemoglobinopathies (6.73%) and β-thalassemia trait (4.86%). The higher prevalence in our study could be due to the selection of only antenatal women. An effort was made to compare with similar studies.

HbD Punjab was the second commonest hemoglobinopathy detected in the present analysis followed by Hb S. HbD Punjab syndromes are not uncommon and are relatively under-diagnosed with heterozygous patients being asymptomatic whereas compound heterozygous states like HbS/D and HbD/beta-thalassemia present with chronic hemolytic anemia requiring frequent red cell transfusions.

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During this present analysis, it was observed that beta thal trait patients had more anemia and low MCV and MCH as compared to Hb D Punjab and Hb S. The anemia was microcytic hypochromic [Table 2]. The findings were comparable to Khera et al., Srinivas et al., and Dhawale et al[10-12]. Therefore, simple routine hemograms along with peripheral blood smear examinations can give us significant clues in diagnosing hemoglobinopathies.

Moreover, public awareness along with various preventive measures may help in the reduction of disease burden. We present this study as additional data about hemoglobinopathies in the reproductive age group female population of Punjab state. We also emphasize that premarital screening should be opted as the mode of a preventive measure as antenatal detection of hemoglobinopathies generates physical and emotional stress for the individual, couple as well as family.

Conclusions:

Educating the carrier female about the potential risk of various hemoglobinopathies may help in controlling the disease. In this study, we targeted the antenatal females, which are the potential source of amplification of disease magnitude and educating them and their family may have a better impact on controlling the birth of child with thalassemia or sickle cell disease.

Ethics approval and consent to participate:

Ethical clearance for the study was obtained from Institutional review board (IRB), Smt. N.H.L. Municipal Medical College, SVP hospital, Ahmedabad.

Conflicts of Interest:

The authors declare that there are no conflicts of interest regarding the publication of this study.

Authors' contributions:

KP analyzed and interpretated the patient data regarding the hemoglobinopathies. GB provided literature material related to this and guidance for the publication.KM and SG helped in typing and collecting necessary informations regarding to the article.NJ helped and guided us in all aspects of article.

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REFERENCES:

1. Weatherall DJ. Postgraduate Hematology. Hemoglobin and the Inherited Disorders of Globin Synthesis. 5th ed. Vol. 2. Oxford Blackwell Science; 2005. p. 245-55.
2. Modell B, Darlison M. Global epidemiology of haemoglobin disorders and derived service indicators. Bull World Health Organ 2008;86:480-7.
3. Madan N, Sharma S, Sood SK, Colah R, Bhatia HM. Frequency of b –thalassemia trait and other hemoglobinopathies in northern and western India. Indian J Hum Genet 2010;16:16-25.
4. Richard A. McPherson, Matthew R. Pincus. Henry's Clinical Diagnosis and Management by laboratory methods. 21st ed. Saunders; 2008. p. 519-532.
5. Philip J, Sarkar RS, Kushwaha N. Microcytic hypochromic anemia: Should high performance liquid chromatography be used routinely for screening anemic and antenatal patients?. Indian J Pathol Microbiol 2013; 56:109-13.
6. Mohanty D, Colah RB, Gorakshakar AC, Patel RZ, Master DC, Mahanta J, et al. Prevalence of β -thalassemia and other haemoglobinopathies in six cities in India: A multicentre study. J Community Genet 2013;4:33-42.
7. Sharma A, Uppal N, Kukreja S, Kaur M, Kaur S. Screening of thalassemia in pregnant females visiting tertiary hospital in Amritsar. Int J Clin Biochem Res 2020;7:226-31.
8. Bhukhanvala DS, Sorathiya SM, Sawant P, Colah R, Ghosh K, Gupte SC. Antenatal screening for identification of couples for prenatal diagnosis of severe hemoglobinopathies in surat, South gujarat. J Obstet Gynaecol India 2013;63:123-7.
9. Narang V, Jain A, Grover S, Soni A, Narang M, Taneja A. Prevalence and spectrum of hemoglobinopathies in females of reproductive age group-A first tertiary care center experience in Punjab, North India. Indian J Patho Microbiol 2023;66:564-7.
10. KheraR,SinghT,KhuanaN,GuptaN,DubeyAP.HPLCincharacterizationof hemoglobin profile in thalassemia syndromes and hemoglobinopathies: A clinicohematological correlation. Indian J Hematol Blood Transfus 2015;31:110-5.
11. Srinivas U, Pati HP, Saxena R. Hemoglobin D-Punjab syndromesin India: A single center experience on cation-exchange high performance liquid chromatography Hematology 2010;15:178-81.
12. Dhawle M, Methwani A, Tangde A, Bindu R. HBQ-India: An uncommon hemoglobin variant. Int J Res Med Sci 2018;6:1687.