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HEMOGLOBINOPATHIES: INSIGHTS FROM A FIVE-YEAR STUDY AT VIVEKANANDA KENDRA NRL HOSPITAL

Pathology

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ABSTRACT

Background: Hemoglobinopathies are major inherited disorders of hemoglobin structure, function, or production, with significant prevalence in the Indian subcontinent. Though they continue to pose a major health concern, early detection and treatment goes a long way in managing these cases. **Objective:** To determine the spectrum and prevalence of hemoglobinopathies in patients attending Vivekananda Kendra NRL Hospital, Numaligarh, Golaghat, Assam, over a five-year period. **Methods:** A retrospective observational study was conducted on 2083 cases between January 2019 and July 2024. Hemoglobin variants were identified using high-performance liquid chromatography (HPLC) with Bio-Rad D10 and Variant analyzers. Demographic, clinical, and hematological datas were analyzed. **Results:** Of the total cases, 1112 (53.4%) demonstrated abnormal hemoglobin patterns. A female predominance (71.5%) was observed, with the highest incidence between 19–50 years. The most prevalent abnormality was HbE trait (47.8%), followed by HbE/β-thalassemia (24.4%), β-thalassemia trait (10.9%), and HbE disease (9%). Sickle cell trait was present in 2.3%, sickle cell with β-thalassemia in 1.4%, and sickle cell disease in 0.16%. One case of hereditary persistence of fetal hemoglobin (HPFH) was documented. **Conclusion:** Hemoglobinopathies are highly prevalent in this region, particularly HbE trait and HbE/β-thalassemia, with significant female predominance. HPLC is a reliable diagnostic tool for large-scale screening. Early detection and awareness programs are essential to reduce the disease burden.

KEYWORDS

INTRODUCTION

Hemoglobinopathies are a group of genetic disorders affecting the quantity or quality of globin chain synthesis. Globally, their prevalence is estimated at 4.2%, with high frequency in the Indian subcontinent, particularly among indigenous populations where thalassemia and sickle cell anemia are common. These disorders can present as asymptomatic traits, moderate anemia, or severe transfusion-dependent conditions. Understanding the regional spectrum of hemoglobinopathies is essential for guiding preventive strategies, genetic counseling, and resource allocation. This study analyzes hemoglobin variants diagnosed at Vivekananda Kendra NRL Hospital over a five-year period.

MATERIALS AND METHODS

The present study is retrospective, cross-sectional study carried out on 2083 patients who were screened by BioRad D10 analyser for hemoglobinopathies at the VKNRL hospital referred from different departments for a period of 5 years from January 2019- July 2024. Although no absolute exclusion criteria were used but sampling was deferred for at least 4 weeks after or just before next transfusion in patients requiring blood transfusions. Hemoglobin variants were identified based on retention times and area percentages. Sickling tests were performed when S-window peaks were detected. Hemoglobin indices and clinical data were also recorded.

RESULTS

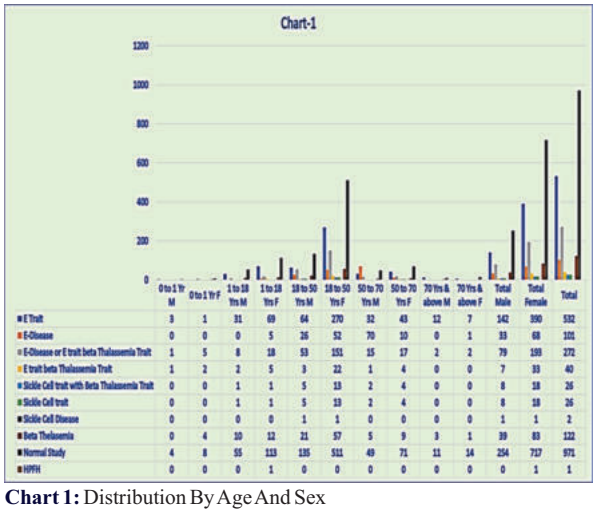

Chart 1: Distribution By Age And Sex

A total of 2083 cases were screened for hemoglobinopathies for 5 years of which 570 were males and 1513 are females. Our study shows female predominance with highest incidence between 19–50 years. Out of 2083 cases, 971 (46.6%) were normal, while 1112 (53.4%) had hemoglobin abnormalities with abnormal hemoglobin showing more prevalence in females 796 (71.5%) compared to males 316 (28.4%). Major variants detected were HbE trait – 532 cases (47.8%), followed by HbE/β-thalassemia – 272 cases (24.4%), β-thalassemia trait – 122 cases (10.9%) and HbE disease – 101 cases (9%). Other abnormalities were also detected which include HbE with β-thalassemia trait – 40 (3.3%) cases,Sickle cell trait – 26 (2.3%)cases, Sickle cell with β-thalassemia – 16 (1.4%)cases, Sickle cell disease 2 (0.16%),HPFH 1 case.

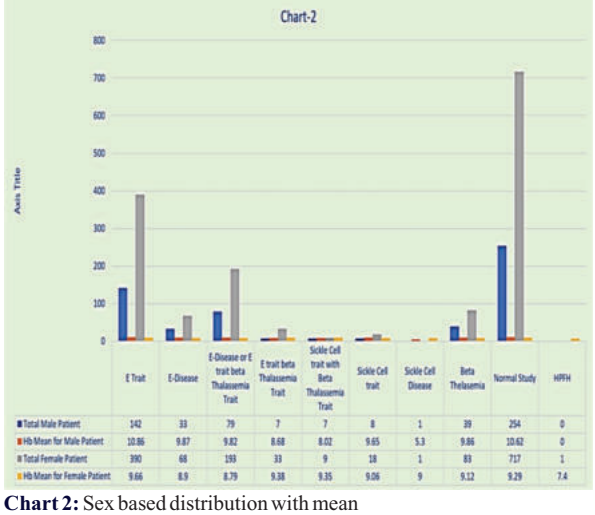

Chart 2: Sex based distribution with mean

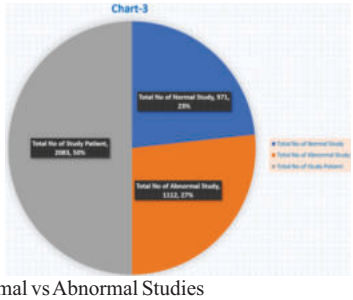

Chart 3: Normal vs Abnormal Studies

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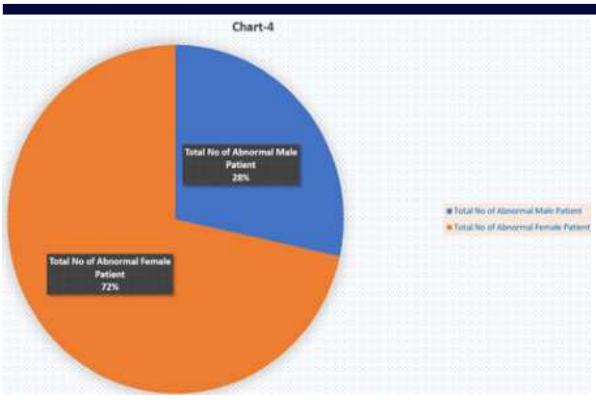

Chart 4: Abnormal studies-gender distribution

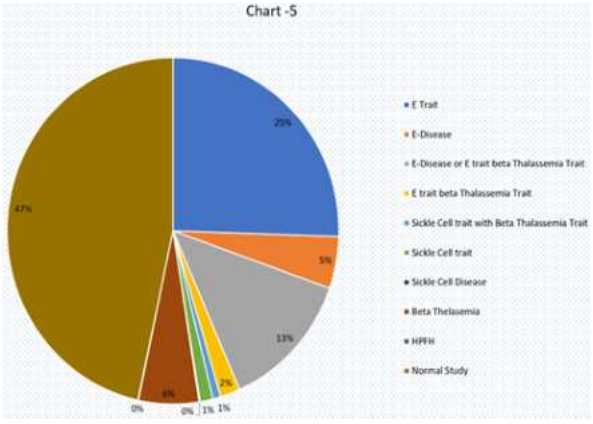

Chart 5: Detailed Distribution Of Abnormalities

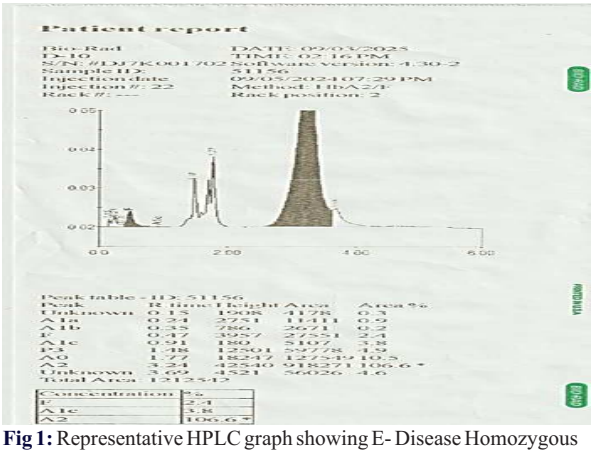

Fig 1: Representative HPLC graph showing E- Disease Homozygous

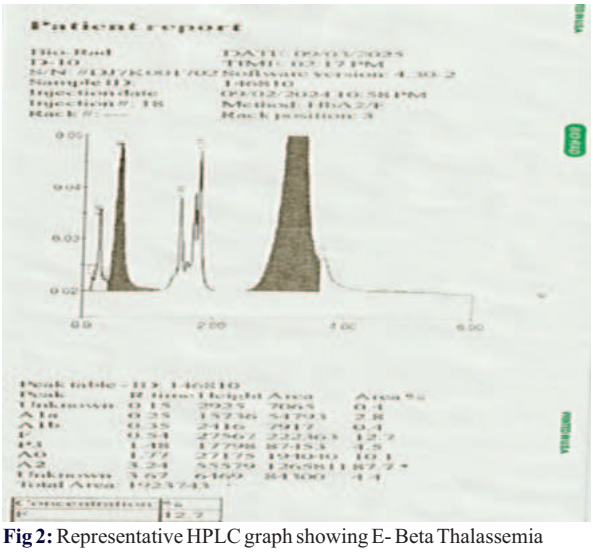

Fig 2: Representative HPLC graph showing E- Beta Thalassemia

DISCUSSION

The present study reveals a high prevalence of HbE trait (47.8%), which is consistent with findings from Bangladesh (Hasan et al., 2023)[1] found in their study 46.02% among 1971 samples. But northern India shows higher case reports of β-thalassemia trait predominates(8.95) according to the study of Sachdev et al[2]. whereas Dolai et al[3] found in their study β-thalassemia trait (10.38%) and HbE trait (4.30%). This indicates that although β-thalassemia trait seems to be the most prevalent hemoglobinopathy in Northern India, our investigation revealed that the prevalence of HbE trait was somewhat higher than that of β-thalassemia type[234]. Our results indicate that HbE/β-thalassemia (24.4%) is a significant health concern in this region, higher than the 1.44% prevalence reported in Dibrugarh (Mohanty et al., 2013)[5]. While HbE carriers are often asymptomatic, HbE/β-thalassemia can present with persistent anemia, ranging from mild to severe[6]. Sickle cell disorders, although less prevalent (2.3% trait, 0.16% disease), still contribute to morbidity[7]. The study also identified one rare case of HPFH, highlighting the spectrum of variants in the population.

CONCLUSION

High-Performance Liquid Chromatography (HPLC) has proven to be a rapid, sensitive, and reliable tool for the screening and diagnosis of hemoglobinopathies. Its utility is particularly significant in the early identification of affected individuals, thereby playing a pivotal role in prevention programs, genetic counseling, and the long-term reduction of healthcare burdens. In this region, the high prevalence of hemoglobinopathies—particularly HbE trait and HbE/β-thalassemia—highlights the need for targeted public health interventions. The observed female predominance and high frequency among young adults underscore potential social and reproductive health implications. Therefore, comprehensive screening and awareness programs are essential to prevent severe forms of the disease and to alleviate the associated economic and psychological stress on affected families. Strengthening collaboration among healthcare providers, governmental agencies, and non-governmental organizations is crucial to enhance preventive strategies and improve health outcomes in populations at risk.

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