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Pathology

SPECTRUM OF SICKLE CELL VARIANTS DIAGNOSED BY HIGH PERFORMANCE LIQUID CHROMATOGRAPHY.

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ABSTRACT **Objective:** Haemoglobinopathies are the most common genetically inherited disorders. Sickle cell disease and its variants result from an abnormal hemoglobin, hemoglobin S (HbS) caused by a single point mutation in Beta-globin gene. Early diagnosis is required so that the next generation can be safeguard against these diseases. Nowadays, HPLC is the most common diagnostic procedure for hemoglobinopathies in India as it is more accurate and rapid. **Method:** This is a prospective observational study conducted in the Department of Pathology, SMIMER Hospital, Surat, Gujarat who had sickle solubility test positive or patients with family history or with abnormal smear findings. **Results:** This study highlights the diversity of sickle cell variants and importance of accurate diagnosis for effective management. **Conclusion:** Cation exchange HPLC is emerging as one of the best methods for screening and detection of various haemoglobinopathies with rapid, reproducible and precise results.

KEYWORDS : HPLC, Sickle cell variants, hemoglobinopathies

INTRODUCTION

Haemoglobinopathies are the most common genetically inherited disorders. Sickle cell disease and its variants result from an abnormal hemoglobin, hemoglobin S (HbS) caused by a single point mutation in Beta-globin gene.

Their prevalence varies with geographical regions. India is developing country and many studies have shown a significant burden of hemoglobinopathies in India. (Vandana Pathak, sharma, & Akshat Agrawal, 2024)

According to World Health Organization (WHO), approximately 5% of world's population carries trait genes for hemoglobin disorders, mainly, sickle cell disease and thalassemia. (Vandana Pathak, sharma, & Akshat Agrawal, 2024)

Early diagnosis of these diseases is required so that the next generation can be safeguard against these diseases. For this purpose, various methods can be used like gel electrophoresis and HPLC. Nowadays, HPLC is the most common diagnostic procedure for hemoglobinopathies in India as it is more accurate and rapid. (Vandana Pathak, sharma, & Akshat Agrawal, 2024)

AIM AND OBJECTIVE

Aim: To investigate the spectrum of sickle cell variants using BIO-RAD (D-10 $HbFA_2/A_1c$) HPLC and to evaluate its diagnostic utility.

Objective:

1) To determine incidence of sickle cell disease according to age and sex group.
2) To classify sickle cell variants based on BIO-RAD (D-10 $HbFA_2/A_1c$) HPLC findings.

MATERIAL AND METHODS

Study Setting- The study was conducted at SMIMER (tertiary care center), Surat, Gujarat, India.

Study Design- Prospective observational study.

Data Collection Period- One year from January 2024 to December 2024.

Inclusion Criteria:

1)All solubility positive samples received from OUT and IN- patient department of SMIMER hospital.
2)All samples suspected for hemoglobinopathies (family history) and those with abnormal peripheral smear findings.

Exclusion Criteria:

Patients with history of blood transfusion within 3 months.

RESULTS AND DISCUSSION

Table -1: Prevalence Of Sickle Cell Variant Hemoglobinopathies In Study Population:

Normal	Sickle cell Anemia	Sickle Cell Trait	Sickle-Beta Thalessemia	TOTAL
235	40 (10%)	84 (22%)	11 (2.9%)	370 (63.5%)

Total 370 samples were screened for hemoglobinopathies.135 were found to have sickle cell variants and 235 showed normal HPLC pattern. 84 (22%) cases were diagnosed as sickle cell trait, 40 (10 %) cases as sickle cell anemia, 11 (2.9%) cases were diagnosed as sickle beta thalassemia.

Among the sickle cell variants, sickle cell trait has been found to be most common. Our study with 84 cases (22%), while it was seen in 3.1% cases done by Pathak V et al.

Table-2: Age Wise Distribution Of Patients.

Age Group	Sickle cell Anemia	Sickle Cell Trait	Sickle-Beta Thalessemia	TOTAL
0-1 years	3	2	4	9
2-10 years	20	4	6	30
11-15 years	5	35	1	41
>15 years	12	43	0	55
Total cases	40	84	11	135

In adult population, 43 cases (78.1%) were of sickle cell trait and 12 cases (21.8%) were of sickle cell disease but no cases were of sickle-beta thalassemia.

Among pediatric age group, patients aged between 0-1 years showed predominance in sickle beta-thalassemia while in patients aged 2-10 years old, sickle cell anemia showed more predominance than sickle cell trait as compared to patients aged between 11-15 years.

97.0% patients of sickle cell disease were below the age of 40. 81.0% patients of sickle cell trait were below the age of 40. 100% patients of sickle beta thalassemia were below the age of 40.

Table-3 Gender Distribution Of Patients:

Sex	Samples
Male	41
Female	94
Total	135

However, Sickle cell trait is more common in our study. When compared among different age groups, SCD showed more predominance than SCT in pediatric age group. In our study the most common age group affected with SCD are from 11-15 years, while it was found in 13-20 years in a study of sickle cell disease by

hematological parameter and HPLC at tertiary care centre. International Journal of Clinical and Diagnostic Pathology by Dr. Mansee et.al. (Chabhadiya, Patel, & Krish, 2024)

In both adult and paediatric age group, female (70%) predominance was more than males (30%). The number of females are high because they are advised HPLC during their antenatal visit.

Gender distribution has shown more female predominance which is about 94 cases (70%) in our study, while it was found to be in 139 cases (61.5%) in a study done by Pathak V et al. (Vandana Pathak, sharma, & Akshat Agrawal, 2024)

CONCLUSION

• Routine screenings, including antenatal and premarital testing, and public health education can mitigate the disease burden.
• Routine sickle solubility screening tests are helpful in early diagnosis and further HPLC of those solubility sickle positive tests are helpful in differentiating sickle cell variants.
• High-Performance Liquid Chromatography (HPLC) is a reliable, precise, and reproducible diagnostic tool for hemoglobinopathies.
• Early diagnosis through HPLC enables accurate classification of variants like HbS, HbA2, and HbF, reducing morbidity and mortality. A premarital counselling can also be done on basis of HPLC results so that the disease may not occur in child. (Vandana Pathak, sharma, & Akshat Agrawal, 2024)

REFERENCES

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