



PREVALENCE OF THALASSEMIA IN TRIBAL PATIENTS ATTENDING TERTIARY CARE HOSPITAL IN JHARKHAND

Pathology

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ABSTRACT

Introduction: The inherited disorders of blood include hemoglobinopathies as one of the major public health problems in India. They cause a high degree of morbidity, moderate to severe hemolytic anemia among vulnerable segments of the society like infants and children, adolescent girls, pregnant women, etc. and several deaths in India. The cumulative gene frequency of the three most predominant abnormal hemoglobins, i.e. sickle hemoglobin, hemoglobin D and hemoglobin E has been found to be 5.35% in India. Prevalence of sickle gene is found to be 0-18% in North eastern India, 0-33.5% in Western India, 22.5-44.4% in Central India and 1-40% in Southern India. The sickling disorders- HbSS, HbSD, HbSE, HbS/ β -thalassemia and other compound heterozygous hemoglobinopathies are all clinically significant, as these combinations present with different manifestations and degrees of severity, making precise identification important. Automated cation-exchange High Performance Liquid Chromatography (HPLC) has emerged as an excellent screening tool for diagnosing these abnormal hemoglobins/ thalassemic states.

- **Aims and Objective:** To determine the prevalence of different types of thalassemias and its distribution in tribal population attending the Pathology Department of RIMS Ranchi.
- To correlate clinical and hematological features of thalassemias.

Material and Methods: The present study entitled "Prevalence of Thalassemia in Tribal patients using High Performance Liquid Chromatography" was carried out in the Department of Pathology, Rajendra institute of Medical Sciences, Ranchi, for a period of one year (Jan 2015-Dec2015). The study was approved by Institutional Ethics Committee, Rajendra Institute of Medical Sciences.

Conclusion: Out of 100 patients diagnosed with thalassemia using HPLC, 28 cases belonged to the Tribal population. In the present study the prevalence of β thalassemia trait in Tribals in Jharkhand was found to be 11%. β Thalassemia major in Tribals in Jharkhand was found to be 7%. Sickle β Thalassemia in Tribals in Jharkhand was found to be 10%. No cases of HbE β Thalassemia in Tribals were found in Jharkhand during this period. The definite identification of disorders of hemoglobin synthesis can be achieved only by DNA analysis, but, in the Indian scenario, family studies on HPLC might be useful as there is paucity of funds, and facilities for DNA analysis are not readily available. Family study is an equally efficacious and cost effective tool. This highlights the role of premarital counseling.

KEYWORDS

HPLC, RETENTION TIME

INTRODUCTION

Thalassemia is considered the most common genetic disorder worldwide. It occurs in a particularly high frequency in a broad belt extending from the Mediterranean basin through the Middle East, Indian subcontinent, Burma and Southeast Asia.

The Scheduled Tribe (ST) population of Jharkhand State is as per 2001 census 7,087,068 constituting 26.3 per cent of the total population (26,945,829) of the State. The Scheduled Tribes are primarily rural as 91.7 per cent of them reside in villages. District wise distribution of ST population shows that Gumla district has the highest proportion of STs (68.4 per cent). The STs constitute more than half of the total population in Lohardaga and Pashchimi Singhbhum districts whereas Ranchi and Pakur districts have 41.8 – 44.6 per cent tribal population. Kodarma district (0.8 per cent) preceded by Chatra (3.8 per cent) has the lowest proportion of the STs Population. Jharkhand has 32 tribal groups:

Munda, Santhal, Oraon, Kharia, Gond, Kol, Kanwar, Savar, Asur, Baiga, Banjara, Bathudi, Bedia, Binjhia, Birhor, Birjia, Chero, Chick-Baraik, Gorait, Ho, Karmali, Kharwar, Khond, Kisan, Kora, Korwa, Lohra, Mahli, Mal-Paharia, Parhaiya, Sauria-Paharia, Bhumij.

The tribes are vulnerable to many hereditary disorders including thalassemia. These genetic disorders adversely affect the general health of an individual. Concerted efforts are therefore required to identify their health issues.¹

MATERIALS AND METHODS

The present study entitled "Prevalence of Thalassemia in Tribal patients using High Performance Liquid Chromatography" was carried out in the Department of Pathology, Rajendra institute of Medical Sciences, Ranchi, for a period of one year (Jan2015-Dec2015). The study was approved by Institutional Ethics Committee, Rajendra Institute of Medical Sciences, Ranchi.

Inclusion Criteria

All patients diagnosed to have thalassemia based on High Performance Liquid Chromatography (HPLC) were included in the study.

It includes β -thalassemia trait, β -thalassemia major, double heterozygous conditions like Sickle- β thalassemia, HbE- β thalassemia and HbD- β thalassemia.

Exclusion Criteria

All patients with inconclusive HPLC results were excluded from the study. The patients having recent blood transfusions: HPLC will not be able to distinguish between patients own cells and transfused cells.

Method used

Cation- exchange HPLC is the method of choice for the initial screening of Hb variants and for accurate quantification of Hb A₂ and HbF concentrations. Bio-Rad Variant II (Bio-Rad Laboratories) is an automated cation exchange HPLC instrument was used to quantify HbA₂, Hb F, HbA along with screening haemoglobin variants like HbS, HbD, HbE and HbC in a single, highly reproducible system.

Guidelines for the Interpretation of results

- 1) The VARIANT II must pass calibration. The retention time for HbA₂ in the calibrator is 3.65±0.10. The calibration factors for haemoglobins A₂ and F must be >0.7 and <1.30, respectively.
- 2) The total area of each analysis should range from 1.0 million to 3.0 million μ volt per second. The results should not be reported if the area is outside this range.
- 3) Quality control values should be in range.
- 4) The normal adult range for HbA₂ is typically 1.75-3.25% of the total haemoglobin. Heterozygous β thalassemia conditions yield HbA₂ levels of 4-9%.
- 5) The normal adult range for HbF is typically <1% of the total haemoglobin. Heterozygous and homozygous conditions of β thalassemia yield HbF levels of 1-5% and 80-100%, respectively.
- 6) P2 and P3 represent minor peaks of hemoglobin A. Levels upto 6% are acceptable.

Analyte Identification Windows

Analyte identification “windows” are intended to assist the laboratory in the interpretation of interpretation of normal and abnormal hemoglobin detected in patient samples. The “windows” are established time ranges in which common variants have been observed to elute using the VARIANT II β -thalassemia Short Program.

The retention time is the centre of the window. Retention time is measured from the time of sample injection to the maximum point of each peak. The band is the half-width of the window.

Analyte Name	Retention Time (Minutes)	Band (Minutes)	Window (Minutes)
F	1.10	0.12	0.98-1.22
P2	1.39	0.11	1.28-1.50
P3	1.70	0.20	1.50-1.90
A0	2.50	0.60	1.90-3.10
A2	3.60	0.30	3.30-3.90
D-WINDOW	4.10	0.20	3.90-4.30
S-WINDOW	4.50	0.20	4.30-4.70
C-WINDOW	5.10	0.20	4.90-5.30

RESULTS

Study Design : A cross sectional study with 100 patients is undertaken to study the prevalence of Thalassemia in tribal patients using High Performance liquid Chromatography.

1. Age distribution of patients :

Age	Tribal Patients	Non Tribal Patients	Total number of Patients	%	% Tribal Patients
<1 yr	1	5	6	6.00%	1.00%
1-10yrs	6	20	26	26.00%	6.00%
11-20yrs	6	15	21	21.00%	6.00%
21-30yrs	11	15	26	26.00%	11.00%
31-40yrs	3	16	19	19.00%	3.00%
40>yrs	1	1	2	2.00%	1.00%
Total	28	72	100	100.00%	28.00%

2. Diagnosis based on HPLC

Name of Haemoglobinopathy	No of Patients(Tribal +Non Tribal)	No of Patients (Tribal)	% prevalence in tribals
Thalassemia β Major	24	7	7%
Thalassemia β Trait	46	13	13%
Thalassemia Intermedia	0	0	0.0%
HbS with β Thalassemia	16	8	8%
HbE with β Thalassemia	14	0	0.0%
HbD with β Thalassemia	0	0	0.0%
Total	N=100	N=28	

3. Distribution of Thalassemia Syndrome in Tribals

Name of Haemoglobinopathy	No of Patients(Tribals)	%
Thalassemia β Major	7	25%
Thalassemia β Trait	13	46.43%
Thalassemia Intermedia	0	0.0%
HbS with β Thalassemia	8	28.57%
HbE with β Thalassemia	0	0.0%
HbD with β Thalassemia	0	0.0%
Total	28	

Hemogram in β Thalassemia in Tribals(n=28)

Name of Disorder	No of Cases	Hb concentration(gm/dl)	MCV (fl)	RDW (%)	HbF (%)	HbA (%)
β Thal Major	7	4.4	71.36	27.08	77.76	17.17
β Thal Trait	11	10.3	78.69	18.38	0.4	84.97

Average Hemogram in mixed haemoglobinopathies in Tribals (n=28)

Name of disorder	No of Cases	Hb concentration (gm/dl)	MCV (fl)	RDW (%)	% of abnormal Hb	HbF(%)
HbS with β Thal	10	7.6	78.65	21.62	19.39	72.25
HbE with β Thal	Nil	Nil	Nil	Nil	Nil	Nil

DISCUSSION:

Distribution of Thalassemia Syndrome in various studies

Type	Balgir RS ²	Rahman et al ³	Shivashankara et al ⁴	Ray Chaudhary S et al ⁵	Present study
Thalassemia trait	18.2%	-	40%	9.62%	11%
Thalassemia major	5.3%	29%	30%	0.47%	7%
Thalassemia intermedia	-	-	-	0.25%	-
HbS with β thalassemia	1.7%	-	-	0.95%	10%
HbE with β thalassemia	0.7%	67%	-	3.67%	-

- A multicentric study was undertaken in six cities of six states of India (Maharashtra, Gujarat, West Bengal, Assam, Karnataka and Punjab) by D.Mohanty, R.B.Colah et al in 2012. The prevalence of β thalassemia trait in Tribals was found out to be:⁶
- 1.4 % in Bangalore, 4.3 % in Kolkata, 2.2% in Mumbai

In the present study the prevalence of β thalassemia trait in Tribals in Jharkhand was found to be 11%.

Conclusion: Out of 100 patients diagnosed with thalassemia using HPLC, 28 cases belonged to the Tribal population. In the present study the prevalence of β thalassemia trait in Tribals in Jharkhand was found to be 11%. β Thalassemia major in Tribals in Jharkhand was found to be 7%. Sickle β Thalassemia in Tribals in Jharkhand was found to be 10%

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