

# Haemoglobinopathies and β-Thalassaemia among the Tribals Working in the Tea Gardens of Assam, India

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## ABSTRACT

**Introduction:** Prevalence of haemoglobinopathies and  $\beta$ -thalassaemia are very high in India but information about its status among the tribals working in the tea gardens of Assam is very less.

Aim: The present study was carried out to determine the prevalence of haemoglobinopathies and  $\beta$ -thalassaemia among the tribals working in the tea gardens of Assam.

**Materials and Methods:** A total 1204 samples from the tribals working in tea gardens of Assam were analysed for both Complete Blood Count (CBC) and High Pressure Liquid Chromatography (HPLC) for detection of haemoglobinopathies and  $\beta$ -thalassaemia.

**Results:** This study showed that the prevalence of sickle cell anaemia and  $\beta$ -thalassaemia were very high among this

population. Our results indicated a higher prevalence of  $\beta$ -thalassaemia (3.07%) among the Munda ethnic group and higher prevalence of sickle cell anaemia (4.73%) among the Lohar ethnic group. This was the first study to report the presence of HbE among the tribals working in the tea gardens of Assam.

**Conclusion:** Based on the present findings, sickle cell anaemia and  $\beta$ -thalassaemia were major health problem for the tribals working in the tea gardens of Assam. Proper diagnostic facilities for haemoglobinopathy and thalassaemia should be established in these areas, including establishment of haemoglobinopathy and thalassaemia database collection, haematological analysis laboratories, genetic counselling clinics, prenatal diagnosis centres and neonatal screening centres.

## Keywords: Haemoglobin E, High pressure liquid chromatography, Prevalence, Sickle cell anaemia

## INTRODUCTION

The haemoglobinopathies are a group of inherited disorders of haemoglobin synthesis. India has the highest concentration of tribal populations globally [1]. Haemoglobinopathy and  $\beta$ -thalassaemia are important health challenges for the tribal populations in India [1]. The sickle cell haemoglobin (HbS), haemoglobin E (HbE) and β-thalassaemia genes are variably distributed across these tribal populations of India. Because of high consanguinity, caste and area endogamy some communities show high rate of incidence making the disease a major public and genetic health problem in India [2,3]. In Assam, the tea garden tribal population constitutes approximately 1/5<sup>th</sup> of the population [4]. Tribals working in the tea gardens of North-East India show HbS as the predominant haemoglobinopathy [5]. In India, HbS has more extensive distribution (10-40% trait frequency) and the homozygotes and double heterozygotes present with a wide array of morbidities [6]. The prevalence of sickle cell carriers among different tribal groups varies from 1-40% [7]. β-thalassaemia and HbE are also prevalent among some tribal groups of India. The prevalence of β-thalassaemia ranges from 6-14% in some tribal populations of Gujarat and Odisha [8,9]. HbE is mainly prevalent among the tribals of North-East, West Bengal, Odisha and Andaman and Nicobar islands [6]. Reports have also showed the prevalence of Hb E/ $\beta$  thalassaemia (2.33% and 1.26%) and Hb S/ $\beta$ -thalassaemia (0.42% and 0.59%) in Assam [10,11]. With migration for work and some intermixing over the years, co-inheritance of HbS with HbD Punjab, HbE and HbC has also been occasionally reported [12-14].

Since very minimal information available on the distribution of  $\beta$ -thalassaemia and other haemoglobinopathies among the different ethnic groups of tribals working in the tea gardens of Assam, the present study was planned to determine the prevalence of haemoglobinopathies and  $\beta$ -thalassaemia among the tribals working in the tea gardens of Assam.

#### MATERIALS AND METHODS

A cross-sectional study was conducted from January 2014 to December 2015 in the tea gardens of six districts (Tinsukia, Dibrugarh, Sivasagar, Jorhat, Golaghat and Dhemaji) of Assam. With 95% confidence interval and at 3.5% margin of error the required minimum sample size is 784. Taking 30% non-response rate and rounding up, the sample size becomes 1200. During the study, total 1204 numbers of samples were collected. These districts have the highest numbers of tea gardens of Assam. From each district randomly four tea gardens were selected. For sample collection health camps were organized in Tinsukia, Dibrugarh and Sivasagar districts. In the other three districts, the concerned doctors of the respective tea gardens helped to collect the samples. To make the residents of the selected tea gardens aware of the survey, letters were handed to the managers and doctors of all the selected tea gardens by the interviewers explaining the objectives of the study. Before sample collection, the interviewers explained about the objectives of the study to the participants. From each tea garden average 50 samples were collected. The age distribution of the study population was between 1 to 70 years. Blood samples were collected after taking informed consent from all the participants. Parental consent was taken in case of children below 16 years of age. A well-designed self-structured and validated proforma was used to get information on the ethnic group, any family history of blood disorders as well as to record all the laboratory findings. The study was approved by the Institutional Ethics Committee.

Patients who had received red blood cell transfusions in the 3 months prior to sample collection were excluded from the study.

All the participants received haematological screening for the presence of  $\beta$ -thalassaemia and other haemoglobinopathies in the Healthcare Clinical Biochemistry Laboratory of Assam Medical College & Hospital, Assam, India. After taking informed consent, 4 ml of peripheral blood was collected in K3EDTA vial from each participant. The samples were collected maintaining proper aseptic

conditions. After collection, samples were transported to the laboratory in insulated packaging by maintaining refrigerated (+2°C to +8°C) condition with the help of gel packs. Complete Blood Count (CBC) was done in cell counter (SYSMEX XS- 800i, Japan) using standard procedure. High Pressure Liquid Chromatography (HPLC, Bio-Rad D10) was performed for all the collected samples.

## RESULTS

A total of 1204 numbers of blood samples were collected and analysed. The whole study population consisted of tribals working in different tea gardens of the region. The distribution of haemo-globinopathies and  $\beta$ -thalassaemia among these ethnic groups; including the origin of these groups were shown in [Table/Fig-1]. The

		*Haemoglobinopathy and Thalassaemia										
Ethnic			HPFH with	Hb S/β-					β-thalassaemia	β-thalassaemia	Hb E/β-	
Groups	Origin of these groups	HbSS	S-window	thalassaemia	Hb SE	Hb AS	Hb EE	Hb AE	trait	major	thalassaemia	Iotal
Bakti	Dunich	0	0	0	0	0	0	0	1	0	0	3
Baraik	Ribar Ibarkband West	0	0	0	0	1	0	0	0	0	0	1
Daraik	Bengal, Port Blair, Odisha	0	0	0	0	1	0	0	0	0	0	
Barhai	Odisha, Bihar	0	0	0	0	1	0	0	1	0	0	2
Bauri	West Bengal	0	0	0	0	2	0	0	4	2	0	8
Bhakta	Odisha	0	0	0	0	0	1	1	1	0	2	5
Bhumij	West Bengal, Odisha, Jharkhand	0	0	0	0	5	0	0	2	0	0	7
Bhuyan	Odisha	0	0	0	0	0	0	1	2	0	0	3
Chick baraik	Bihar, Jharkhand, West Bengal, Port Blair, Odisha	6	0	1	0	17	0	1	3	0	0	28
Chowtal	Odisha	0	0	0	0	0	0	0	1	0	0	1
Doom	Odisha	0	0	0	0	0	0	1	0	0	0	1
Ganju	Jharkhand	0	0	0	0	0	0	0	2	0	0	2
Garh	Chhattisgarh	8	0	0	0	15	0	0	0	0	0	23
Ghatowar	Odisha	1	0	0	0	4	0	0	0	0	0	5
Gonds	Madhya Pradesh, Chhattisgarh, Andhra Pradesh, Odisha	0	0	0	0	0	0	1	0	0	0	1
Guwala	Uttar Pradesh	1	0	2	0	8	0	0	4	0	0	15
Kamar	Chhattisgarh	0	0	0	0	1	0	0	0	0	0	1
Kandha	Odisha	0	0	0	0	0	0	0	1	0	0	1
Karua	Tamil Nadu	0	0	0	0	1	0	0	1	0	0	2
Kashyap	Uttar Pradesh	1	0	0	0	5	0	0	0	0	0	6
Kumari	Telangana	0	0	1	0	0	0	0	0	1	0	2
Kunda	Uttar Pradesh	1	0	0	0	0	0	0	1	0	0	2
Kurmi	Uttar Pradesh	0	0	0	0	2	0	0	4	0	0	6
Lohar	Bihar	16	0	2	0	39	0	2	7	1	1	68
Mahali	West Bengal, Odisha	1	0	0	0	3	0	0	0	0	0	4
Majhwar	Uttar Pradesh	0	0	0	0	2	0	0	0	0	0	2
Mali	North India, East India	0	0	0	0	1	0	0	0	0	0	1
Manjhi	Uttar Pradesh	0	0	0	1	1	0	0	6	1	0	9
Munda	Jharkhand,Odisha, West Bengal, Chhattishgarh, Bihar	1	0	0	0	25	2	5	31	5	1	70
Nayak	Karnataka	3	0	0	0	10	0	0	1	0	0	14
Oraon	Jharkhand, Chhattisgarh, West Bengal, Odisha, Bihar	0	1	0	0	1	0	1	10	3	0	16
Panika	Chhattisgarh, Madhya Pradesh, Odisha, Andhra Pradesh	0	0	2	0	2	0	0	0	0	0	4
Parja	Odisha	0	0	0	0	1	0	0	2	1	0	4
Patnayak	Odisha	4	0	0	0	5	0	0	0	0	0	9
Pator	Odisha, Bihar, Jharkhand	1	0	0	0	6	0	0	1	0	0	8
Sahu	Bihar, Jharkhand	0	0	1	0	1	0	0	1	0	0	3
Saora	Odisha, Andhra Pradesh, Jharkhand, Madhya Pradesh	0	0	1	0	3	1	0	6	0	0	11
Tanti	Bihar, West Bengal	14	1	6	1	29	0	0	9	1	0	61
Tasa	Bihar	1	0	0	0	4	0	0	0	0	0	5
Teli	Bihar, Jharkhand	0	0	0	0	3	0	0	1	0	0	4
Total		59	2	16	2	199	4	14	104	15	4	419

[Table/Fig-1]: Distribution of Haemoglobinopathies and β-thalassaemia in different ethnic groups of tribals working in the tea gardens of Assam. \*HbSS: HbS homozygous, HPFH with S-window: Hereditary Persistance of Foetal Haemoglobin with S window, Hb S/β- thalassaemia: Compound HbS /β- thalassaemia, Hb SE: Compound heterozygote HbS/HbE, HbAS: HbS trait, Hb EE: HbE homozygous, HbAE: HbE trait, HbE/β-thalassaemia: Compound HbE/β-thalassaemia prevalence of  $\beta$ - thalassaemia was highest among the Munda ethnic group (3.07%) and the prevalence of HbS was highest among the Lohar ethnic group (4.73%) [Table/Fig-1].

During the study, haemoglobinopathies and  $\beta$ -thalassaemia were detected in 34.8% of the participants [Table/Fig-2]. The prevalence of haemoglobinopathies and  $\beta$ -thalassaemia found in the study population were shown in [Table/Fig-2]. The distribution of haemoglobinopathies and  $\beta$ -thalassaemia among different age groups were shown in [Table/Fig-3]. The age grouping for the study was done following WHO standard [15]. 1.66% of the participants were compound heterozygotes of  $\beta$ -thalassaemia which co-inherited

Variables <sup>#</sup>	Case Number	Percentage (%)
Hb AA	785	65.2
Hb SS	59	4.9
HPFH with S-window	2	0.17
Hb S/β- thalassaemia	16	1.33
Hb SE	2	0.17
Hb AS	199	16.53
Hb EE	4	0.33
Hb AE	14	1.16
β-thalassaemia trait	104	8.64
β-thalassaemia major	15	1.25
Hb E/β-thalassaemia	4	0.33

[Table/Fig-2]: Prevalence of Haemoglobinopathies and  $\beta$ -thalassaemia in the study population.

\*HbAA: Normal, HbSS: HbS homozygous, HPFH with S-window: Hereditary Persistance of Foetal Haemoglobin with S-window, Hb S/β- thalassaemia: Compound HbS/β-thalassaemia, HbSE: Compound heterozygote HbS/HbE, HbAS: HbS trait, HbEE: HbE homozygous, HbAE: HbE trait, Hb E/β-thalassaemia: Compound HbE/ β- thalassaemia with HbE and HbS. During the study 0.33% of homozygous HbE (HbEE) and 1.16% of HbE Trait (HbAE) cases were detected. Within the study population 27% cases had severe anaemia, 22% had mild anaemia and 39% had moderate anaemia. The data also revealed that the literacy rate was very low in this population [Table/Fig-4]. Within the whole population 30.23% were illiterate.

## **DISCUSSION**

The high prevalence rate of haemoglobinopathy and  $\beta$ -thalassaemia could cause serious health and social problems among the tribals working in the tea gardens of the region. Preventing the birth of affected children is the best possible option for India to control haemoglobinopathies and  $\beta$ -thalassaemia [16]. A prerequisite for this is the knowledge of the prevalence of  $\beta$ -thalassaemia and other haemoglobinopathies in different regions of the country and particularly in different ethnic groups [16]. In India, marriages are usually arranged among individuals of the same caste or ethnic groups and so it is important to find out the prevalence of B-thalassaemia and also HbE among the different ethnic groups [16]. The current study was carried out to detect the prevalence of  $\beta$ -thalassaemia and haemoglobinopathies among the tribals working in the tea gardens of Assam based on their ethnicity. This study population is a distinct occupational group who migrated from states like Madhya Pradesh, Bihar, Orissa and Andhra Pradesh to work as tealeaf picker and have been residing within the tea-estates of the region [17,18]. The general incidence of β-thalassaemia trait and sickle cell anaemia varies between 3-17% and 1-44%, respectively in India [2,3]. The average sickle cell gene frequency was found to be highest 9.1% in Orissa, 7.4% in Madhya Pradesh, 7.2% in Uttar Pradesh and 7.1% in Tamil Nadu [19]. In upper Assam, the HbS gene frequency was 5.11% [11]. In India, the prevalence of Sickle cell trait (HbAS) varies from 5-40% among tribal populations

	Haemoglobinopathies and Thalassaemia											
Age Group	Hb AA	Hb SS	HPFH with S-Window	Hb S/β- thalassaemia	Hb SE	Hb AS	Hb EE	Hb AE	β-thalassaemia trait	β-thalassaemia major	Hb E/β- thalassaemia	Total
0-4	8	2	0	1	1	5	0	1	3	7	0	28
5-9	8	15	0	1	0	5	0	0	3	5	4	41
10-14	65	9	0	1	0	13	1	1	5	2	0	97
15-19	117	16	0	4	0	19	0	2	16	1	0	175
20-24	209	7	1	5	1	38	0	4	16	0	0	281
25-29	146	6	1	2	0	36	2	3	20	0	0	216
30-34	66	1	0	0	0	22	0	1	11	0	0	101
35-39	79	2	0	0	0	25	0	1	9	0	0	116
40-44	28	0	0	0	0	9	0	1	10	0	0	48
45-49	31	0	0	0	0	15	0	0	4	0	0	50
50-54	12	1	0	0	0	7	0	0	3	0	0	23
55-59	7	0	0	0	0	2	1	0	4	0	0	14
60-64	5	0	0	0	0	3	0	0	0	0	0	8
65-69	2	0	0	2	0	0	0	0	0	0	0	4
70-74	2	0	0	0	0	0	0	0	0	0	0	2
Total	785	59	2	16	2	199	4	14	104	15	4	1204

[Table/Fig-3]: Prevalence of haemoglobinopathies and  $\beta$ -thalassaemia among different age groups.

	Haemoglobinopathies and Thalassaemia												
Education	Hb AA	Hb SS	HPFH with S-Window	Hb S/β- thalassaemia	Hb SE	Hb AS	Hb EE	Hb AE	β-thalassaemia trait	β-thalassaemia major	Hb E/β- thalassaemia	Total	
Illiterate	227	18	0	5	1	60	1	3	34	12	3	364	30.23%
Primary & middle	366	24	2	6	1	74	1	7	32	3	1	517	42.94%
Secondary	134	15	0	5	0	44	2	4	25	0	0	229	19.02%
College & above	58	2	0	0	0	21	0	0	13	0	0	94	7.81%
Total	785	59	2	16	2	199	4	14	104	15	4	1204	100%
[Table/Fig-4]: Literacy rate among the study population.													

from different states [20]. Among the four tribal communities of Visakhapatnam, HbAS distribution was 1.69% among Konds, 14.36% among Bagatas, 7.8% among Konda Doras and 13.59% among Konda Kammaras [20]. In Chhattisgarh, the prevalence of HbAS was 9.3% [21]. Studies done among the tribal groups in Orissa showed the prevalence rate of  $\beta$ -thalassaemia trait from 6.3% to 8.5% [22,23]. In the present study, 16.53% cases have sickle cell trait and 8.64% have β-thalassaemia trait [Table/Fig-2]. Another study from Orissa reported that the prevalence of HbAS was 5.3% among Kharia and Bhuyan tribe and the prevalence of β-thalassaemia trait was 6.2% in Kharia and 6.6% in Bhuyan [24]. The present study has shown that the prevalence of β-thalassaemia and sickle cell anaemia were highest among the Munda and Lohar respectively. According to our knowledge, this is the first study to report the presence of Hb E among the tribals working in the tea gardens of Assam [Table/Fig-1].

This study was also designed to provide information and awareness among the tribals working in the tea gardens about the genetic risks of having haemoglobinopathies and  $\beta$ -thalassaemia.

### LIMITATION

The present study has limitations as the study sites have not covered the whole state of Assam.

#### CONCLUSION

This study showed that sickle cell anaemia and  $\beta$ -thalassaemia were major health problem for the tribals working in the tea gardens of Assam. Therefore, the local government and the health departments have to develop a system of prevention and control program to decrease haemoglobinopathies and thalassaemia in this population. We suggested that prevention network of thalassemia should be established in this region, including establishment of a thalassemia database, haematological analysis laboratories, genetic counselling clinics, prenatal diagnosis centres and neonatal screening centres. Research in these areas should continue focusing on various challenges in care delivery, prevention and basic sciences on interaction of haemoglobinopathies with various other infections.

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