



## Haemoglobinopathies in Saharia: A particularly vulnerable tribal group of Shivpuri district of Madhya Pradesh

### KEYWORDS

Haemoglobinopathies, hematology, sickle cell anaemia, thalassaemia, Saharia, Madhya Pradesh

### BP Urade

Nandini Bhattacharyya, CS Roy and Joy Das

#### ABSTRACT

Haemoglobinopathies are caused by abnormalities in haemoglobin molecule. Sickle cell anaemia (SCA), thalassaemia (Hb $\beta^T$ ) and other variants of haemoglobin attribute to a major health problem in India. Of the several haemoglobinopathic disorders, HbS, Hb $\beta^T$ , HbE and HbD are widely prevalent in Indian sub-continent. For the present study, 2 ml intravenous blood samples were drawn from 309 (113 males and 196 females) Saharia PVTG inhabits in extremely remote area in Shivpuri district of Madhya Pradesh. Capillary electrophoresis of 309 blood samples showed 17 individuals (13 females and 4 males) positive for haemoglobinopathies out of which 3 individuals found for sickle cell trait (SCT) and 14 for  $\beta$ -thalassaemia (Hb $\beta^T$ ). That means 5.5 per cent of the Saharia population possess these two abnormal hemoglobin genes. The result shows higher frequency of  $\beta$ -thalassaemia (4.53 per cent) than sickle cell anaemia (0.97 per cent) among the Saharia, a PVTG of Madhya Pradesh. This is a unique situation unlike any other tribal communities where the predominant of  $\beta$ -thalassaemia exists among the Saharia PVTG of Madhya Pradesh but the haplotype that is found among other communities is same or differs can only be ascertained at the molecular level.

Hematologic components such as WBC, Hb, HCT, MCV, MCH, MCHC, and PLT show higher mean values in normal individuals compared to Hb $\beta^T$  individuals except RBC and RDW. The MCH and MCHC components were considerably low in Hb $\beta^T$  individuals as expected. Since no data on sickle cell is available among the Saharia, the present study would pave the way to go in to deeper aspect of it so as to arrive at concrete conclusion on whether the HbS gene has got its unicentric origin by independent mutation or infiltrated.

To prevent and manage this disorder from spreading further in future generation, detection of this genetic abnormality through premarital age group and prenatal diagnosis is must and the government should make these tests mandatory and effectively for entire population.

#### INTRODUCTION

Haemoglobinopathies are group of monogenic disorders those are concerned with the abnormality in the protein molecules of Red Blood Cells. A genetic defect that results in abnormal structure of one of the globin chains of the haemoglobin molecule is termed as haemoglobinopathy. The genetic defect producing structural abnormalities in the haemoglobin is due to substitution of one amino acid for another (e.g. HbS and HbE). The other form of genetic defect is produced functionally deficient globin chains what is called thalassaemia. Beta thalassaemias ( $\beta$ -thal) are a group of inherited blood disorders which reduce the production of hemoglobin. These forms of thalassaemia are caused by reduced or absent synthesis of the beta chains of hemoglobin that result in variable outcomes ranging from severe anemia to clinically asymptomatic individuals. Due to this situation a person's body makes an abnormal form of haemoglobin which is the protein molecule in red blood cells which carries oxygen. Very few studies have been conducted especially on PVTG's (Feroz, 2001; Debnath, 2014; Chakma et al., 2006; Urade, 2012c; Mohanty et al., 2015).

Among various haemoglobinopathy disorders, sickle cell anaemia is the most common inherited blood disease. According to WHO, 5 per cent of the world's population carries trait genes for haemoglobinopathy disorder such as thalassaemia and sickle cell anaemia.

#### MATERIAL AND METHOD

A two days Health Awareness Camp for 'Community genetics at High Risk Zones for Thalassaemia, Sickle Cell Anaemia' under the project -Community Genetics and Health: Bio-cultural Adaptation was held at Kakrai village in Pohari Block of Shivpuri district of Madhya Pradesh on 15<sup>th</sup> and 16<sup>th</sup> September, 2015. During camp 2 ml intravenous blood samples were collected in B. D. vacutainer (USA) from 309 individuals comprising 113 males and 196 females and brought to Survey's DNA lab at CRC, Nagpur. Data has been collected from 28 villages. Haematological parameters were analysed for all the blood samples using automated Coulter machine. All the blood samples were subjected to Sebia capillary electrophoresis for different haemoglobin variants. The status and pattern of sickle cell and  $\beta$ -thalassaemia were noted down according to the pattern of peak and cut-off per cent (>3.5%) in a given zone. DNA has been

extracted from all the positive samples for further analysis at molecular level.

#### RESULTS AND DISCUSSION

Table - 1 shows sex-wise distribution of sickle cell trait and  $\beta$ -thal among the Saharia population. Of the 309 Saharia individuals, 17 of them showed (13 females and 4 males) haemoglobin variants of HbS and Hb $\beta$ -thal. Out of 17 individuals, 3 were found for HbS and 14 for  $\beta$ -thal. It means that Hb $\beta$ -thal is more frequent than the sickle cell gene among the Saharia, a PVTG of Madhya Pradesh. Only 5.5 per cent of the Saharia population possesses these two types of haemoglobin variants comprising of sickle cell trait and  $\beta$ -thalassaemia i.e., 0.97 per cent and 4.53 per cent respectively. It means that the Saharia a PVTG have both these genes. On the contrary, earlier studies reported by Debnath (2014) and Chakma et al. (2006) did not find sickle cell gene among the Saharia except the presence of  $\beta$ -thalassaemia.

The results of haematological parameters for normal individuals and carriers of HbS and  $\beta$ -thalassaemia are shown in Table - 2. Less value of MCV, MCH, and MCHC in  $\beta$ -thalassaemia indicates microcytic hypochromic anaemia results from impaired haemoglobin synthesis.

Different haematological aspect between normal and Hb $\beta$ -thal carriers have been depicted in Fig. 1. The comparison of different haematological parameters shows that mean RBC and RDW of  $\beta$ -thal carriers were higher than their normal counterparts. As expected the mean of MCV, MCH, and MCHC which are prime indicators to determine the status of  $\beta$ -thal show lesser values than the normal individuals. Likewise, mean of WBC, Hb, HCT, and PLT were also higher values in normal individuals than the  $\beta$ -thal carriers.

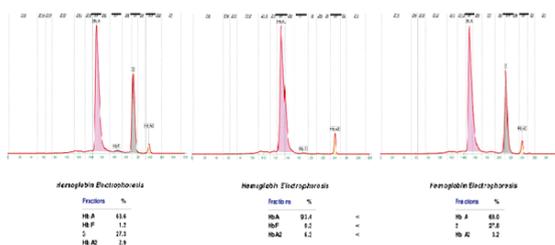
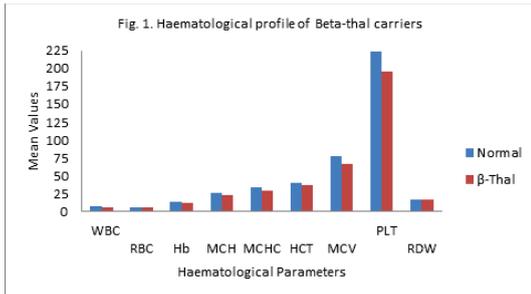
**Table - 1. Phenotypes of Sickle cell and  $\beta$ -thal among the Saharia of Shivpuri district**

	Sickle cell Anaemia			$\beta$ -thal	
	AA	AS	SS	Carrier	Affected
M	109	-	-	4	-
F	183	3	-	10	-
%	94.5	0.97	-	4.53	-

**Table- 2** Mean values of Haematological parameters

Normal Individuals	WBC	RBC	Hb	HCT	MCV	MCH	MCHC	PLT	RDW
Mean	7.49	5.24	13.65	40.29	77.33	26.21	33.78	222.83	16.88
SD (±)	5.52	0.75	2.16	5.68	8.89	3.70	1.64	97.94	2.47
-thal	6.50	5.46	12.60	37.21	65.86	22.33	29.17	195.51	17.01
SD (±)	1.39	0.73	4.73	14.22	25.48	8.31	12.16	58.39	1.38

- Feroz, M and K Aravindan 2001: Sickle cell disease in Wayanad, Kerala: gene frequencies and disease characteristics. *Natl Med J India*, 14:267 – 270.
- Mohanty, D, MB Mukherjee, RB Colah, M Wadia, K Ghosh, GP Chottray et al. 2015: Spectrum of hemoglobinopathies among te primitive tribes: A multicentric study in India. *Asia Pac J Public Health*, 27: NP 562 – 571.
- Urade, BP 2012c: Marital distance and genetic structure among the Mullukuruman of Kerala. *J Anthropol Sur India*, Vol 26(1):211-222.



**CONCLUSION**

Due to lack of access to health education and basic health facilities the tribal people are highly vulnerable to genetic and communicable diseases owing to poverty and ignorance. The infiltration of β-thalassaemia in the Saharia, a PVTG in Madhya Pradesh is unique one unlike other Indian tribal populations and PVTG's. The low frequency of sickle cell gene compared to β-thalassaemia in the Saharia characterized by the geographical and genetic isolation. Some authors have reported complete absent of sickle cell gene among the Saharia but β-thalassaemia is prevalent. The present study found the prevalence of sickle cell gene though at the lesser degree (0.97 per cent) compared to β-thalassaemia (4.53 per cent) suggested that the Saharia, is not free from these two types of haemoglobin variants.

In the recent past there has been a trend in the rise of number of people becoming victim of these dreaded genetic disorders. The prevailing situation has led the governments and regulatory authority to initiate awareness programme to decrease the burden of haemoglobinopathies and control the mortality rate out of it. To prevent and manage these disorders from spreading further in future generation, detection of this genetic abnormality through premarital age group and prenatal diagnosis followed by genetic counselling is must and the government should make these tests mandatory and effectively for entire population.

**Acknowledgement**

Authors are grateful to the Director, Anthropological Survey of India, Kolkata for sanctioning this project, Shri J Sreenath for his untiring co-operation throughout study, Shri Sanjay Shukla, Shri Khisti, Shri S. Maske, Ms. Rupali Mange for their assistance and all the Saharia people who voluntarily donated blood samples.

**References:**

- Chakma, T, P VinayRao, PK Meshram, SB Singh 2006: Health and nutritional profile of tribals of Madhya Pradesh and Chhattisgarh. *Proceedings of National Symposium on Tribal Health*, Jabalpur, RMRCT.
- Debnath, D. 2014 : The health problems and status of the particularly vulnerable tribal groups of Madhya Pradesh – A case study in Anthropological dimension, *Man in India*, 94(4):597 – 609