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The prevalence of sickle cell disease phenotypes and sickle cell gene frequency in some tribals of Melghat forest region of Amravati, Maharashtra (India)

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ABSTRACT :
Sickle cell disease (SCD) is a major genetic disorder among the tribal population. Hence the objective of the present study was to determine the prevalence and frequency of the sickle cell gene in some selected tribal population of the Melghat forest region in Amravati district (Central India). A total of 438 tribal individuals were screened for SCD from 06 tribal villages constituting 05 tribal castes (Korku, Bhil, Gaoli, Gowari and Nihal). Using electrophoresis on cellulose acetate membrane 43 individuals were found to be heterozygous and 12 individuals were found to be homozygous for sickle cell gene. The Sickle cell allele frequency was found to be 0.3294 in Korku, 0.4934 in Bhills, 0.4071 in Gaoli, 0.2871 in Gowari and 0.2898 in Nihal.

Keywords: *Sickle cell anaemia, Tribals, Korku, Bhil, Melghat.*

INTRODUCTION

Sickle cell disease (SCD) is an autosomal recessive genetically transmitted haemoglobinopathy responsible for considerable morbidity and mortality. This is hereditary blood disorder due to defective haemoglobin structure. Sickle cell disorder is caused by a point mutation at 6th position in beta globin chain, valine substituting glutamic acid due to which in deoxygenated state the shape of erythrocytes change to sickle shape and also the fragility of cell membrane increases. Prior to 1952, no information was available about the existence of sickle cell disease in India. In 1952 it was recorded for the first time, simultaneously amongst the tribal population group of Nilgiri hills and labourers in the tea garden of Assam. Now it is firmly established that these genes harbor amongst different caste groups but with very high prevalence amongst scheduled castes, scheduled tribes and other backward communities (Baig *et al.,* 2004).

The prevalence of sickle cell gene has been reported in many parts of India including Central India, where the prevalence in different communities ranges from 9.4% to 22.2% (Shukla & Solanki 1985). Available data indicates that SCD gene is widely spread in all districts of eastern Maharashtra (Vidarbha region), Northern Maharashtra (Satpuda range) and some parts of Marathwada region (Sharma 1983, Shukla & Solanki 1985.) Population genetic survey data has shown that the overall prevalence of sickle cell disorder in different tribal population in the state of Maharashtra is 10% for carriers state and 0.5% for the sufferers (Kate 2001). The prevalence is very high amongst the tribal population groups from Nandurbar and Gadchiroli district of the state. Kate (2001) reported SCD prevalence of 10% among the Korku tribes of Amravati district. However, no detail investigation has been done to study the prevalence of sickle cell disorder in the different tribal groups of Amravati district. Hence in the present work an effort has been made to screen few tribal hamlets of Melghat forest region, Amravati district and find out the magnitude of the prevalence of SCD in the tribal groups residing in this region.

MATERIAL AND METHODS :-

Screening of SCD was conducted in 06 tribal villages of Amravati district from September 2009 to 2010 and total of 438 blood samples from individuals belonging to 05 different tribal castes were collected by door to door screening or by organizing screening camps. The screening of the tribal village population was performed by taking into confidence the police patil of the village and the villagers were approached through him for making them aware of the screening camp. Few drops of blood were collected by bold finger prick for performing the solubility test for preliminary diagnosis of SCD. Blood samples of solubility test positive subjects were later subjected to electrophoresis on cellulose acetate membrane (Dacie & Lewis, 1991) as a confirmatory test for SCD, in the pathology laboratory at Govt. Medical College, Nagpur.

Allele frequency was calculated using Hardy-Weinberg principle. Coefficients of gene differentiation (G_{ST}) and (D_{ST}) were calculated following Nei (1973). The genetic relationship among the present endogamous tribal population groups were assessed using the measure of

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genetic distance (D) proposed by Nei (1972). A dendrogram was drawn as per UPGMA clustering method using Phylip (v 3.69). (Felsensteins, 1993).

RESULTS AND DISCUSSION

The allele frequency of sickle cell gene was found to be highest in the Bhil tribal group followed by Gaoli, Korku and Nihal and was lowest in the Gowari (Table – 1). The SS genotype frequency was found to be highest in Korkus and lowest in the Bhils (Figure – 1). The AS genotype frequency was found to be highest in the Korkus and lowest in the Gowari. The chi-square differences in sickle cell allele frequency for different tribal population was found to be significant in Korku and Bhil population whereas nonsignificant in the Goali, Gowari and Nihal population (Table – 2).

The gene diversity indices for total populations H_T and intratribal population gene diversity (H_S) was quite high i.e. 0.901 and 0.801 respectively (Table - 3). The interpopulation gene diversity (D_{ST}) is also 1.00 and the coefficient of genetic differentiation (G_{ST}) is 0.667.

The dendrogram constructed using genetic distance obtained from the sickle gene allele frequencies using UPGMA method shows two main embranchment or clade designated as A and B. The clade A exhibits clustering of Bhil with the Gaoli while clade B shows clustering of Gowari with the Nihal. Interestingly, all these four endogamous population belong to the Indo-Aryan group. Of note, the korku,a tribe belonging to the Austroasiatic linguistic group occupies basal position in the dendrogram. Moreover, the status of Austroasiatic tribal as the earliest settler of Indian subcontinent is well established (Roychoudhury *et al.,* 2000, Baig *et al.,* 2004).

Population	Observed	Phenotype		Allele frequency	
		Sickling test +ve	Sickling test -ve	A	S
Korku (n=212)	No. %	23 10.84	189 89.16	0.6706	0.3294
Bhil (n=53)	No. %	14 24.13	44 75.87	0.5066	0.4934
Gaoli (n=54)	No. %	09 16.66	45 83.34	0.5929	0.4071
Gowari (n=54)	No. %	04 07.41	47 92.59	0.7129	0.2871
Nihal (n=63)	No. %	05 07.93	58 92.07	0.7102	0.2898

Table 1: Solubility test data: Phenotype and Allele frequency

Population	X^2	p-value	df
Korku	0.6974	0.4036	1
Bhil	1.0926	0.2958	1
Gaoli	0.4462	0.5041	1
Gowari	0.0849	0.7706	1
Nihal	0.1075	0.7429	1

Table 2: Values of X^2, p & df

Locus	D_{ST}	H_T	H_S	G_{ST}
Sickle cell gene	1.000	0.901	0.801	0.110

Table 3: Sickele cell gene diversity analysis

	Korku	Bhil	Gaoli	Gowari	Nihal
korku					
bhil	0.024689				
gaoli	0.010565	0.012906			
gowari	0.002721	0.038899	0.024140		
nihal	0.002393	0.037852	0.023132	0.000010	

Table 4: Genetic distance matrix

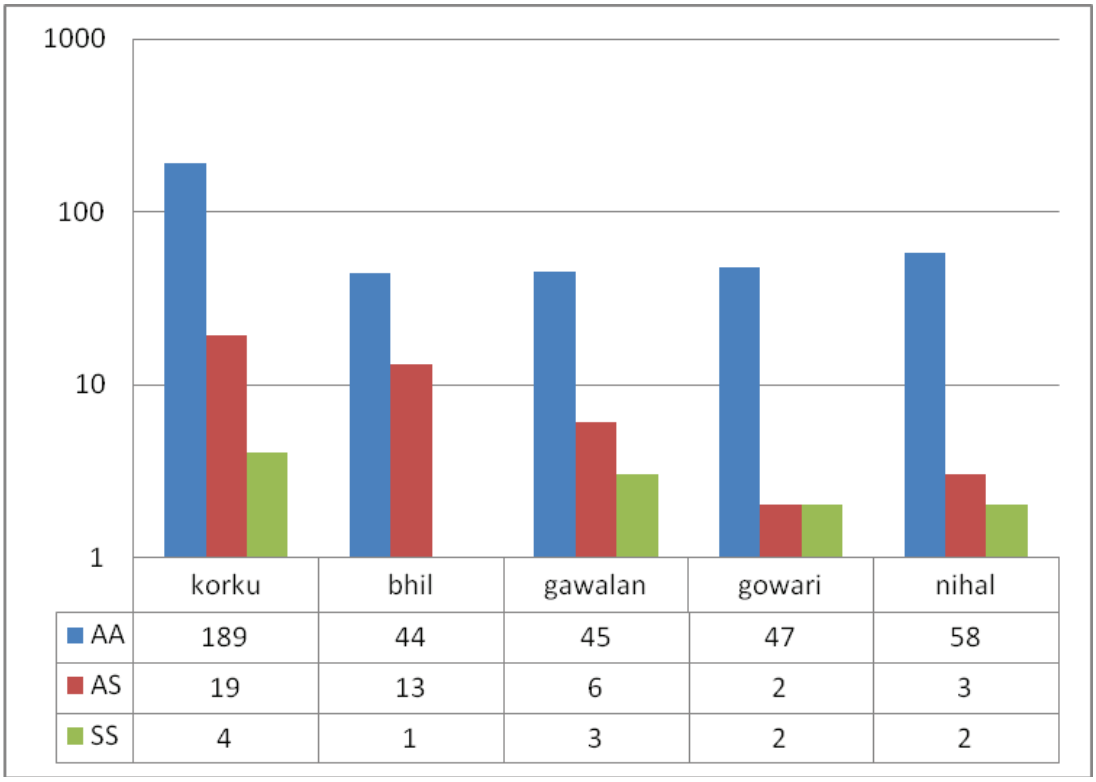

Fig1: Bar Chart showing the distribution of sickle cell phenotypes in 5 tribal endogamous tribal population.

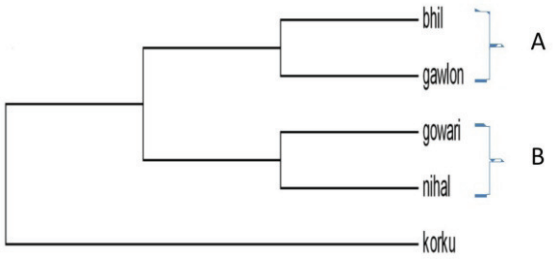

Fig.2: Dendrogram showing genetic relationship among five endogamous populations of Amravati district, Maharashtra.

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