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Investigations on the Variability of Genetic Markers in Three Tribal Populations from Maharashtra, India

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ABSTRACT Three tribal populations (Koli, Konkana, Pawara) from different regions of Maharashtra with a total of n=456 male and female individuals were typed for seven polymorphic blood group (A1A2BO, MNSs, RHESUS; Koli and Konkana only) and serum protein group systems (HP,GC, TF, PI). The genetic variability among these populations is considerable which can be attributed to locally acting differentiation processes and which could be preserved because of their geographical isolation from each other.

INTRODUCTION

Though in the last decades many studies on the variability of genetic markers have been carried out on Indian populations, which were recently summarized and critically discussed by Bhasin, Walter and Danker-Hopfe (1992) as well as by Walter, Danker-Hopfe and Bhasin (1991), there are still many gaps concerning our understanding of the regional and ethnic variability of these markers in India. As for Maharashtra detailed studies have been performed especially by Sanghvi (1978), Mukherjee et al. (1979a, b) and Walter et al. (1992, in press), which revealed a considerable genetic variability among the various populations groups of this state. However, our knowledge on the variability of genetic markers in this part of India is far from being sufficient, and in particular the tribal populations living in Maharashtra were hitherto scarely typed for genetic polymorphisms of the blood.

In order to contribute to our knowledge on the distribution of genetic markers in the tribal populations of Maharashtra we collected blood samples from three different groups-Koli, Konkana and Pawara-and analyzed them for various blood and serum protein groups. The results of our investigations will be presented here in detail.

MATERIAL AND METHODS

We collected blood samples from the following three tribal groups from Maharashtra:

1. Koli: This is tribal community residing in Western Maharashtra, particularly in the districts of Nasik, Thane and Pune. This group is characterized by a very low incidence (0-2.0%) of the sickle cell trait. The Koli are predominantly vegetarians. They worship God Shiva and practise consanguinity.

2. Konkana: This tribal group is located in the Nasik District and in the areas between the borderline of Maharashtra and Gujarat. The incidence of the sickle cell trait in them varies between 6-8%. They eat mixed food, *i.e.*, vegetarian, fish and meat. The Konkana practise consanguinity and do not marry other tribals.

3. Pawara: This tribal group is located in the Dhulia district of Maharashtra. They live in the borderline area of Maharashtra and Madhya Pradesh in the hilly ranges of the Satpara Mountains. The incidence of the sickle cell trait in the Pawara is very high: 25%. They

worship Hindu Gods. The Pawara do not marry to outside tribals and practise consanguinity.

Figure 1 shows the location of these three tribal groups.

Blood samples of a total of n=456 male and female individuals were taken by venipuncture. The A_1A_2BO, MNSs and RHESUS blood groups were typed in the Department of Pediatrics, Medical College and Sasoon General Hospitals, Pune, whereas the serum samples were air-mailed deep-frozen to Bremen, where HP,GC, TF, and PI typings were done in the Department of Human Biology of the University of Bremen. Unfortunately it was not possible to type also the Pawara sample for the blood group polymorphisms.

All the typings were done as described by Spielmann and Kühnl (1982) and Prokop and Göhler (1986).

RESULTS AND DISCUSSION

A_1A_2BO system: Table 1 shows the distribution of A_1A_2BO phenotype and allele frequencies in the Koli and Konkana. Both groups are in genetic equilibrium.

Table 1 : A_1A_2 BO phenotype and allele frequencies in two tribal populations from Maharashtra

	Koli		*Konkana*	
Phenotypes	*Obs.*	*Exp.*	*Obs.*	*Exp.*
A_1	12	12.35	74	75.03
A_2	5	5.14	12	12.15
B	28	28.76	36	36.49
O	31	31.84	40	40.56
A_1B	5	4.04	22	19.47
A_2B	3	1.88	4	4.29
Total	84	84.01	188	187.99
χ^2		0.959		0.379
df		2		2
p		ns		ns
Alleles				
A_1		0.1028		0.2948
A_2		0.0478		0.0650
B		0.2337		0.1757
O		0.6157		0.4645
Total		1.0000		1.0000

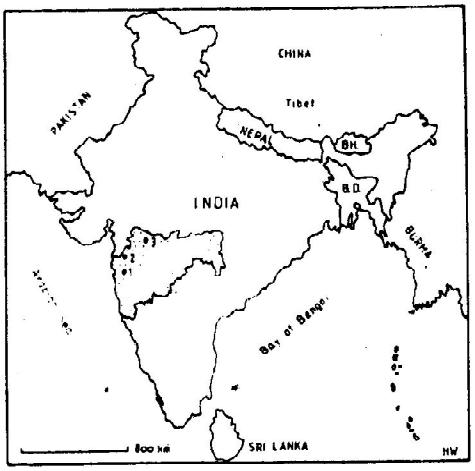

Fig. 1. Location of Maharashtra (dotted). 1=Koli, 2=Konkana, 3=Pawara, B.H.=Bhutan, B.D.=Bangla Desh

Table 2 : MN phenotype and allele frequencies in two tribal populations from Maharashtra

	Koli		*Konkana*	
Phenotypes	*Obs.*	*Exp.*	*Obs.*	*Exp.*
M	30	29.76	48	51.60
MN	40	40.48	101	93.78
N	14	13.76	39	42.61
Total	84	84.00	188	187.99
χ^2		0.012		1.113
df		1		1
p		ns		ns
Alleles				
M		0.5952		0.5239
N		0.4048		0.4761
Total		1.0000		1.0000

Table 3 : RHESUS phenotype and haplotype frequencies in two tribal populations from Maharashtra

	Koli		*Konkana*	
Phenotypes	*Obs.*	*Exp.*	*Obs.*	*Exp.*
CCD.EE	—	0.14	—	0.02
CCD.Ee	4	4.14	3	2.99
CCD.ee	29	30.07	108	108.38
CCddee	1	0.13	—	0.25
CcD.EE	—	0.47	—	0.25
CcD.Ee	8	8.76	18	19.00
ccD.EE	2	0.38	2	0.78
ccD.Ee	2	3.23	3	3.92
CcD.ee	30	27.72	48	45.31
Ccddee	1	1.09	1	1.13
ccD.cc	4	4.58	3	3.71
ccddee	2	2.29	1	1.25
Total	83	83.00	187	186.99
χ^2		14.218		3.065
df		6		6
p		0.05>p>0.02		ns
Haplotypes				
cde		0.1661		0.0818
Cde		0.0394		0.0368
cDe		0.1216		0.0811
CDe		0.5638		0.7254
CDE		0.0414		0.0105
cDE		0.0677		0.0644
Total		1.0000		1.0000

MN system: The distribution of MN phenotype and allele frequencies in the Koli and Konkana is demonstrated in table 2. Genetic equilibrium can be assumed for both of them.

RHESUS system: RHESUS phenotype and haplotype frequencies are shown in table 3. Whereas genetic equilibrium can be assumed for the Konkana, some deviation from it is seen in the Koli group, which, however, is most likely due to sample size.

Table 4 : HP phenotype and allele frequencies in three tribal populations from Maharashtra

	Koli		*Konkana*		*Pawara*	
Phenotypes	*Obs.*	*Exp.*	*Obs.*	*Exp.*	*Obs.*	*Exp.*
HP 1	1	0.66	6	4.10	6	4.32
2-1	14	14.68	45	48.78	37	40.37
2	82	81.66	147	145.12	96	94.31
Total	97	97.00	198	198.00	139	139.00
χ^2		0.208		1.198		0.965
df		1		1		1
p		ns		ns		ns
Alleles						
*HP*1*		0.0825		0.1439		0.1763
*HP*2*		0.9175		0.8561		0.8237
Total		1.0000		1.0000		1.0000

Table 5: GC phenotype and allele frequencies in three tribal populations from Maharashtra

	Koli		*Konkana*		*Pawara*	
Phenotypes	*Obs.*	*Exp.*	*Obs.*	*Exp.*	*Obs.*	*Exp.*
GC 1F	2	1.73	5	4.63	2	2.20
1F-1S	15	14.60	35	35.35	22	20.69
1S	31	30.86	69	67.52	49	48.59
2-1F	7	7.96	16	16.39	10	10.89
2-1S	33	33.67	60	62.61	49	51.16
2	10	9.18	16	14.51	15	13.47
Total	98	98.00	201	201.01	147	147.00
χ^2		0.256		0.336		0.442
df		3		3		3
p		ns		ns		ns
Alleles						
*GC*1F*		0.1327		0.1517		0.1224
*GC*1S*		0.5612		0.5796		0.5749
*GC*2*		0.3061		0.2687		0.3027
Total		1.0000		1.0000		1.0000

HP system: Table 4 shows HP phenotype and allele frequencies in the three tribal groups under study. Genetic equilibrium can be assumed for all of them.

GC system: The distribution of GC phenotype and allele frequencies is demostrated in table 5. Genetic equilibruum is seen in all the groups. Rare GC variants were not recorded.

TF system: Table 6 shows the TF phenotype and allele frequencies in the three tribal groups. No deviation from genetic equilibrium is seen. Rare TF variants were not found.

Table 6 : TF phenotype and allele frequencies in three tribal populations from Maharashtra

	Koli		*Konkana*		*Pawara*	
Phenotypes	*Obs.*	*Exp.*	*Obs.*	*Exp.*	*Obs.*	*Exp.*
TF C 1	68	67.77	100	92.01	78	80.80
2-1	27	27.45	70	85.92	55	50.37
2	3	2.78	28	20.06	6	7.85
3-1	—	—	2	2.04	4	3.01
3-2	—	—	1	0.95	—	0.94
3	—	—	—	0.01	—	0.03
Total	98	98.00	201	200.99	143	143.00
χ^2		0.026		6.801		2.250
df		1		3		3
p		ns		ns		ns
Alleles						
*TF*C1*		0.8316		0.6766		0.7517
*TF*C2*		0.1684		0.3159		0.2343
*TF*C3*		—		0.0075		0.0140
Total		1.0000		1.0000		1.0000

PI system: The distribution of PI phenotype and allele frequencies in the three tribal groups is demonstrated in table 7. Genetic equilibrium can be assumed for the Koli and Konkana group, whereas the Pawara shows some deviation from it. It cannot be concluded that this deviation is also due to chance. Rare PI variants were not observed.

Comparing the allele and haplotype frequencies, respectively, of the three tribal populations under study one can observe considerable differences among them. In order to examine how far this genetic heterogeneity is statistically significant we tested the locus and allele specific heterogeneity according to Rao (1952). The results of this analysis are shown in table 8. For the loci A_1A_2BO, RH, HP, TF and PI a more or less statistical significance if seen, whereas the loci MN and GC do not show significant heterogeneity. One can conclude from this that the differences in the distribution of MN and GC alleles in the three tribal groups are due to chance.

Table 7 : PI phenotype and allele frequencies in three tribal populations from Maharashtra

	Koli		*Konkana*		*Pawara*	
Phenotypes	*Obs.*	*Exp.*	*Obs.*	*Exp.*	*Obs.*	*Exp.*
PI M 1	39	39.23	63	63.23	48	42.72
2-1	40	39.86	62	61.86	50	54.73
2	10	10.12	15	15.13	19	17.53
3-1	6	5.69	43	42.69	7	12.84
3-2	3	2.89	21	20.88	10	8.12
3	—	0.21	7	7.21	3	0.96
Total	98	98.00	211	211.00	137	137.00
χ^2		0.231		0.011		8.561
df		3		3		3
p		ns		ns		0.05>p>0.02
Alleles						
*PI*M1*		0.6327		0.5474		0.5584
*PI*M2*		0.3214		0.2678		0.3577
*PI*M3*		0.0459		0.1848		0.0839
Total		1.0000		1.0000		1.0000

Concerning the A_1A_2BO locus it is seen that the interpopulation heterogeneity is mainly due to the highly significant variation of the *ABO*A1* and *ABO*O* frequencies. The intrapopulation heterogeneity in the RHESUS system is in particular caused by the variation of the haplotypes *cde*, *CDe* and *CDE*, whereas that in the TF system is mainly due to the variation of *TF*C1* and *TF*C3* alleles, and that in the PI system by the variation of *PI*M2* and in particular *PI*M3* alleles. It can be supposed that the genetic heterogeneity among the three tribal populations of Maharashtra is

the result of microevolutionary effects as *e.g.* genetic drift or founder effects. As all the three tribal groups under study are geographically quite distant from each other (*see* Figure 1) one can suppose that they did not have genetic contacts with each other so that they could preserve their specific genetic profiles. Thus one can regard the geographical isolation of these three tribal groups from each other as that factor for the emergence and maintenance of the marked genetic heterogeneity among them. The results of our investigations on these tribal groups are also a further example for the eminent role of geographical isolation concerning genetic differentiation process in man.

Table 8: Locus and allele specific heterogeneity

Locus/Allele		χ^2	*df*	*p*
ABO		25.96	3	***
	A_1	23.78	1	***
	A_2	0.62	1	ns
	B	2.50	1	ns
	O	10.62	1	**
MN		2.38	1	ns
RH		19.57	5	**
	cde	8.50	1	**
	Cde	0.02	1	ns
	cDe	2.22	1	ns
	CDe	13.70	1	***
	CDE	5.60	1	*
	cDE	0.02	1	ns
HP		8.39	2	*
GC		2.15	4	ns
	*GC*1F*	1.28	2	ns
	*GC*1S*	0.18	2	ns
	*GC*2*	1.35	2	ns
TF		19.36	4	***
	*TF*C1*	16.85	2	***
	*TF*C2*	16.14	2	***
	*TF*C3*	2.93	2	ns
PI		32.25	4	***
	*PI*M1*	4.17	4	ns
	*PI*M2*	6.55	2	*
	*PI*M3*	29.56	2	***

*** : p<0.001; ** : 0.01>p>0.001
* : 0.005>p>0.01; ns : p>0.05

Comparing finally the results of this study with those obtained on two other tribal populations from Maharashtra (Maria Gond and Raj Gond from Chandrapur, East Maharashtra) one can point out that the West and East Maharashtrian tribals differ obviously in almost all polymorphic blood group and serum protein systems. This follows from the figures as given below (for details *see* Walter et al., 1992; Walter et al., in press):

This is not the place to discuss these genetic differences in detail, but it should be pointed out here briefly that they also reflect without doubt the role of geographical and hence genetic isolation for the understanding of the intraspecific genetic diversity of man. A detailed analysis and discussion of the genetic heterogeneity of the various population groups of Maharashtra will be given elsewhere.

Allele/Haplotype	*Maria Gond*	*Raj Gond*
A_1	0.1445	0.1938
A_2	0.0417	0.0544
B	0.2706	0.1925
0	0.5432	0.5432
	n= 116	n= 106
M	0.6897	0.4953
N	0.3103	0.5047
	n=116	n=106
cde	0.1083	—
Cde	0.0294	—
cDe	—	0.0928
CDe	0.7718	0.8207
CDE	0.0048	0.0158
cDE	0.0857	0.0707
	n=116	n=104
*HP*1*	0.0804	0.1068
*HP*2*	0.9196	0.8932
	n= 112	n= 103
*GC*1F*	0.1681	0.1571
*GC*1S*	0.6207	0.5809
GC*2	0.2112	0.2571
*GC*Var*	—	0.0049
	n=116	n=105
*PI*M1*	0.4741	0.5524
*PI*M2*	0.3103	0.2524
*PI*M3*	0.2069	0.1857
*PI*Var*	0.0087	0.0095
	n= 116	n= 105

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