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RED CELL ENZYME POLYMORPHISM (ADA, AKI, ESD, PGM1, GPI, ACP1) IN JAT SIKHS AND MUSLIMS OF PUNJAB

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ABSTRACT

The allele frequencies in human populations are used in different areas such as population genetics, forensic genetics and anthropological studies. Various different populations have been investigated their allele frequency distributions of polymorphic traits. The aim of the present genetic study was to provide data on the distribution of various red cell enzyme markers of Jat Sikhs and Muslims of Punjab. Blood samples from two major caste populations Jat Sikhs (1831) and Muslims (992) of Punjab was taken for analysis. The main aim of present study was to provide baseline data for these two groups for a battery of six red cell enzyme polymorphisms (ADA, AK1, ESD, PGM1, GPI, ACP1), following standard biochemical protocols. Results showed that there were appreciable differences among the present castes in the distribution of the first three polymorphic enzymes (ADA, ESD, PGM1) while no significant heterogeneity was discernible in the next three (AK1, GPI, ACP1); some GPI variants have been reported form the state. It was attempt to seek an interpretation of genetic variability of biochemical data in Jat Sikhs and Muslims of Punjab.

Keywords: Enzyme Polymorphism, Caste Populations, Punjab

INTRODUCTION

The frequencies of alleles in human populations reflect the genetic structure of populations and used in establishing the relationship of different populations to each other. Besides, these informations are valuable for the genetic, forensic and

anthropological studies [1, 2]. For investigation of the allele frequency distributions, mainly polymorphic systems such as blood group antigens, leukocyte antigens (HLA), erythrocyte enzymes, serum proteins, hemoglobin variants and STR loci of DNA are used. Up to now a vast amount of data on the allele frequencies of the polymorphic traits has been reported from various populations. During the last over three decades a large number of different human red cell enzyme polymorphisms have been investigated using electrophoretic techniques in a variety of different populations. As far their distributions in world populations, comprehensive compilations have appeared from time to time [3-5]. For people of the Indian region, such data are listed by Bhasin and Singh [6, 7]. Up to now a vast amount of data on the allele frequencies of the polymorphic traits has been reported from various populations [8-17]. The present study was planned to reveal genetic heterogeneity in Jat Sikhs from Majha and Doaba, a ubiquitously present agricultural caste population of Punjab, north-west India and Muslims of Malerkotla town of Sangrur district where nearly 80% of the population is comprised by the Muslims.

MATERIALS AND METHODS

The present genetic study has been restricted to only two caste population of Punjab i.e. the Jat Sikhs and Muslims of Malerkotla. The subjects for this biochemical genetics study comprised a total of 1831 apparently healthy not closely related Jat Sikh subjects born and brought up in Punjab. Out of these 1061 Jat Sikh samples were collected from districts of Gurdaspur, Amritsar (Majha zone), Hoshiarpur, Kapurthala, Jalandhar and Nawanshahr (Doaba zone) and 770 samples from Ferozepur, Muktsar, Bhatinda, Sangrur, Rupnagar (Malwa zone). Blood samples from 992 apparently healthy not closely related Muslim subjects were collected in batches from different schools of Malerkotla. The subjects selected at random were attending various educational institutes in these districts. Blood collections were made in batches, spread over several field works. Haemolysates prepared by freezing and thawing technique, were electrophoresed for studying variabilities of different red cell enzyme in the present study. All haemolysates were freshly typed within weeks for phenotypes of seven, different polymorphic red cell enzymes viz., Adenosine deaminase (ADA), Adenylate kinase locus 1 (AK1), Esterase D (ESD), Phosphoglucomutase locus 1 (PGM1),

Glucosephosphatase isomerase (GPI) and Acid phosphatase locus 1 (ACP1) and by horizontal agarose gel electrophoresis following standard methods. The isozymes of ADA and AK1 were electrophoresed on a single gel and stained simultaneously [18]. Similarly, ESD and PGM1 were also separated together on the same gel and stained in the same order following the technique of Wraxall and Stolorow [19]. For the phenotypings of red cell enzymes GLO1, ACP1 and GPI electrophoresis and stainings were performed with some modifications of the original techniques [20-22]. From phenotype data allele frequencies were calculated using the gene counting method [23]. Deviations from Hardy- Weinberg equilibrium were studied by goodness-of-fit chi square.

RESULTS AND DISCUSSION

Table 1 showed phenotypic distribution of red cell enzyme polymorphism ADA, AK1, ESD, PGM1, GPI, ACP1 in Jat Sikhs and Muslims of Punjab. Table 2 and 3 represented allele frequencies and Goodness of fit Chi square test.

Adenosine Deaminase (ADA)

Data obtained on phenotypes of ADA system and allele frequencies calculated along with goodness of fit Chi-square values among the studied Jat Sikh population groups inhabiting the different districts of Punjab. All the three common phenotypes of the enzyme viz., ADA 1, 1,2 and 2 were found present in Jat Sikhs and Muslims of Punjab but no example of any rare phenotype of the system was detected in these two caste groups (Table 1). ADA*2 frequency was reported to be 0.1249 in Jat Sikhs and 0.1707 in Muslims of Punjab. When the present six Jat Sikh groups from Punjab were compared with Muslim of Punjab statistically, these two sects showed significant differences in this enzyme system also (χ^2= 16.314, d.f. 2).

Adenylate Kinase Locus 1 (AK1)

Both the phenotypes AK1 and AK2 were found to be in both Jat Sikhs and Muslims of Punjab. The *AK1*2* allele frequency in Jat Sikhs was reported to be 0.1132 while *AK1*2* frequencies were observed 0.2182 in Muslims of Punjab respectively (Table 1 and 2). Comparison of the AK1 data of Jat Sikh groups with Muslims of Punjab showed homogeneous distribution of the polymorphism (χ^2 = 0.050, d.f. 2) (Table 3).

Esterase D (ESD)

The frequency of the *ESD*2* allele was observed to be 0.2026 in Jat Sikhs and 0.4427 in Muslims of Punjab. Comparison of all the six studied groups of the caste from north Punjab with Muslims of Punjab also reveal significant heterogeneity in this

enzyme among them (χ^2 = 11.295, d.f. 2) (Table 1-3).

Phosphoglucomutase Locus 1 (PGM1)

Results of electrophoretic typings of erythrocyte enzyme phosphoglucomutase locus 1 (PGM1) in the Jat Sikh population groups studied here from various northern districts of Punjab state showed that *PGM1*2* allele frequency was found to be 0.3043 and 0.5647 in Jat Sikhs and Muslims of Punjab respectively. Rare phenotype *PGM1*7* was reported in Jat Sikhs of Punjab and *PGM1*3* was encountered in Muslims of Punjab. Typing of PGM1 system in the present Muslim sects and Jat Sikhs from Punjab demonstrated that statistical significant differences between the two groups were reported (χ2 = 9.565, d.f. 2) (Table 3). Rare alleles of this enzyme system such as *PGM1*5* [24,25] (Papiha *et al.,* 1976; Roberts *et al.,* 1974) and *PGM1*7* [26-28] have been encountered in the Muslims of India, this is the first report on the presence of the rare allele *PGM1*3* in them.

Glucosephosphate Isomerase (GPI)

In addition to common phenotype GPI 1, rare variant phenotype GPI 3 was found in Jat Sikhs and Muslims of Punjab while variant GPI 7 was encountered only in Jat Sikhs and variant GPI 5 was reported in Muslims of Punjab. Using the phenotype data the frequency of the rare GPI*3 allele was calculated to be 0.0031 in the Jat Sikhs and 0.0039 in Muslims and frequency of rare allele GPI*7 was found to be 0.0003 in Jat Sikhs and GPI*5 was found to be 0.0006 in Muslims of Punjab. Comparison of the GPI data of Jat Sikh groups with Muslims of Punjab showed homogeneous distribution of the polymorphism (χ^2 = 3.902, d.f. 2).

Acid Phosphatase Locus 1 (ACP1)

In the two sects of the present Jat Sikhs and Muslim material from Punjab, all the three common phenotypes of the system (ACP1 A, A,B, B) were present in the Jat Sikhs and Muslims of each studied group. In addition, phenotypes ACP1 A, C and B,C were also present albeit in very low proportions. However, no rare ACP1 phenotype (variant) was encountered in the present genetic survey from Punjab (Table 1 and 2). The frequencies of three common alleles of the system viz. *ACP1*A, ACP1*B* and *ACP1*C* were found to be 0.3401, 0.6535 and 0.0064 in Jat Sikhs and 0.6711, 1.3137 and 0.0151 in Muslims of Punjab (Table 2). In fact, the Chi-square test revealed that both these sects are genetically close (χ2 = 2.521, d.f. 2), so deviations are due to chance only (Table 3).

Table 1: Phenotype distribution of various red cell enzyme systems in the Jat Sikhs and Muslims of Punjab

System	Phenotype	Jat Sikhs (n=1,831)	Expected	Muslims (n==992)	Expected
ADA	ADA1	1394	1433.41	816	776.59
	ADA 1,2	415	380.73	172	206.27
	ADA 2	22	16.86	4	9.14
	χ^2 HW(df=1)	16.314		1.674	
AK1	AK1 1	1440	1439.25	779	779.75
	AK1 1,2	367	367.11	199	198.89
	AK1 2	24	14.44	14	13.35
	χ^2 HW(df=1)	0.050		0.045	
ESD	ESD 1	1163	1129.21	578	611.79
	ESD 1,2	593	612.28	351	331.72
	ESD 2	75	89.51	63	48.49
	χ^2 HW(df=1)	11.295		0.381	
PGM1	PGM1 1	885	914.85	525	495.15
	PGM1 1,2	764	742.91	381	402.09
	PGM1 2	178	169.99	84	92.01
	PGM1 1,3	-	0.65	1	0.35
	PGM1 1,7	4	2.60	-	1.40
	χ^2 HW(df=1)	9.565		0.760	
GPI	GPI 1	1818	1819.97	988	986.03
	GPI 1,3	12	9.73	3	5.27
	GPI 1,5	-	0.65	1	0.35
	GPI 1,7	1	0.65	-	0.35
	χ^2 HW(df=1)	3.902		0.00	
ACP1	ACP1 A	233	237.39	133	128.61
	ACP1 A,B	774	765.35	406	414.65
	ACP1 B	801	808.16	445	437.84
	ACP1 A,C	5	5.19	3	2.81
	ACP1 B,C	18	14.92	5	8.08
	χ^2 HW(df=1)	2.521		3.323	

Table 2: Allele frequencies of enzyme systems investigated in the Jat Sikhs and Muslims of Punjab

System	Allele	Jat Sikhs	Muslims
ADA	*ADA*1*	0.7923	1.8293
	*ADA*2*	0.1249	0.1707
AK1	*AK1*1*	0.8867	1.7818
	*AK1*2*	0.1132	0.2182
ESD	*ESD*1*	0.7973	1.5573
	*ESD*2*	0.2026	0.4427
PGM1	*PGM1*1*	0.6946	1.4327
	*PGM1*2*	0.3043	0.5647
	*PGM1*3*	-	0.0026
	*PGM1*7*	0.0010	-
GPI	*GPI*1*	0.9967	1.9955
	*GPI*3*	0.0031	0.0039
	*GPI*5*	-	0.0006
	*GPI*7*	0.0003	-
ACP1	*ACP1*A*	0.3401	0.6711
	*ACP1*B*	0.6535	1.3137
	*ACP1*C*	0.0064	0.0151

Table 3: Inter-group Chi-square comparisons (d.f. 2) for various erythrocyte enzyme polymorphisms between the Jat Sikh and Muslims of Punjab

Jat Sikh X Muslims	χ^2
ADA	16.314*
AK1	0.050
ESD	11.295*
PGM1	9.565*
GPI	3.902
ACP1	2.521

CONCLUSION

In conclusion, the allele frequencies of Jat Sikhs are closer to Muslims for AK1, GPI, ACP1 while significant heterogeneity was found in three enzymes ADA, ESD, PGM1. Rare variants of GPI 3, 5 and 7 have been reported in at Jat Sikhs and Muslims of Punjab.

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