Original Communication

Red cell enzyme and serum protein polymorphisms (ACP1, PGM1, GLO1, ESD, HP, PI) in Turkish population

L. Dönbak, C. Csete[1], S. Salaçin[2], T. Varga[1]
Kahramanmaras Sütçü Imam University, Science and Arts Faculty, Department of Biology, Kahramanmaras-Turkey, [1]Albert Szent-Györgyi Medical University, Department of Forensic Medicine, Kossuth Lajos Sugarut 40, Szeged, Hungary, [2]Dokuz Eylül University, School of Medicine, Department of Forensic Medicine, Izmir-Turkey

BACKGROUND: 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.
AIMS AND OBJECTIVES: The allele frequency distributions of four red cell enzyme (ACP1, PGM1, GLO1, ESD) and two serum protein systems (HP, PI subtypes) were analyzed in Turkish population from Adana area, Turkey, in order to enlarge our knowledge on the genetic composition of Turkish population.
MATERIAL AND METHODS: Venous blood samples taken from 200 unrelated Turkish individuals were transported to the Forensic Serology Laboratory of Albert Szent-Györgyi Medical University (Szeged/Hungary) for phenotyping. The phenotypes of ACP1, PGM1, GLO1, ESD, and HP systems were determined by means of starch gel electrophoresis, while PI subtypes were obtained by polyacrylamide gel isoelectric focusing method. The allele frequencies were calculated by gene counting.
RESULTS: The calculated frequencies of the alleles are as follows: ACP1*A = 0.250 ± 0.021, ACP1 *B = 0.693 ± 0.023, ACP1 *C = 0.057 ± 0.011; PGM1 *1 = 0.710 ± 0.022, PGM1*2 = 0.288 ± 0.022, PGM1*6 = 0.002 ± 0.002; GLO1*1 = 0.373 ± 0.024, GLO1*2 = 0.627 ± 0.024; ESD*1 = 0.805 ± 0.019, ESD*2 = 0.195 ± 0.019; HP*1 = 0.265 ± 0.022, HP*2 = 0.735 ± 0.022; PI*M1 = 0.887 ± 0.015, PI*M2 = 0.010 ± 0.004, PI*M3 = 0.095 ± 0.014, PI*S = 0.008 ± 0.004.
CONCLUSIONS: The comparison of the data with those of Asian and European populations showed that the allele frequencies of ACP1, ESD and HP are similar or close to those of Asian populations, whereas those of PGM1 and PI come close to European populations. The allele frequency of GLO1 system is inbetween those of Asians and Europeans.

Key words: Erythrocyte enzymes, Serum proteins, Polymorphisms, Population genetics, Turkish population

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 frequeny distributions, mainly polymorphic systems such as blood group antigens, leucocyte 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.[3-12]

In the present study, Acid phosphatase 1 (ACP1), Phosphoglucomutase 1 (PGM1), Glyoxalase 1 (GLO1), Esterase D (ESD), Haptoglobin (HP) phenotypes and Alpha-1-Antitrypsin (PI) subtypes were investigated in Turkish population from Adana area, localized in the eastern Mediterranean region of Turkey, with the purpose of enlarging our knowledge on their genetic constitution.

Materials and Methods

The blood samples were taken from 200 healty, 17-58 years old male and female unrelated volunteer donors. Blood samples collected into tubes containing anticoagulant were transported within a insulated container at +4 °C to the Forensic Serology Laboratory of Forensic Medicine Department, Albert Szent-Györgyi Medical University (Szeged, Hungary) for analysing. Samples were divided into plasma and erythrocyte aliquots. Red cell lysates were prepared by freezing and thawing washed red cells. Plasma samples were treated with hemoglobin solution for HP phenotyping before electrophoresis.

Address for Correspondence: L. Dönbak, Kahramanmaras Sütçü Imam Üniversitesi, Fen Edebiyat Fakültesi, Biyoloji Bölümü, Avsar kampüsü, Kahramanmaras/Türkiye. E-mail: lale@ksu.edu.tr

Starch gel electrophoresis were used to determine the phenotypes of ACP1, PGM1, GLO1, ESD, and HP phenotypes. The subtyping of PI system were performed by isoelectric focusing (IEF) on polyacrylamide gel.[13]

Allele frequencies were calculated simply by gene counting method. The goodness of fit test between the observed values and those expected under the assumption of Hardy-Weinberg equilibrium were performed by using χ^2-test.

Results

The observed and expected phenotype and the allele frequencies of the genetic markers studied are shown in Table 1. The observed phenotype frequencies were in agreement with the Hardy-Weinberg equilibrium expectations in all systems.

No rare electrophoretic variants of the GLO1, ESD and HP systems were detected in this Turkish population sample. However, in the ACP1 system, the CC rare homozygote phenotype was observed in one case. As shown in Table 1, the allele frequency of ACP1*C calculated as 0.057. Besides, among the rare PGM1 variants, only PGM1*6 was encountered with the frequency of 0.002. Furthermore, rare variant, PI*S in the PI system has been observed. The allele frequency of PI*S was found to be 0.008 as seen in Table 1.

Discussion

In order to investigate genetic composition of Turkish population, the allele frequencies of six genetic traits were calculated and compared with those of Asian and European populations.

ACP1

Previous studies indicated that geographic distribution of ACP1*B and ACP1*A alleles show a correlation with climate in Europe. The frequency of ACP1*B increases with the increase in the mean annual temperature. ACP1*B, the most common allele of this system, reach its highest frequency in African and Asian populations.[2] The ACP1*C allele, lack in most of African, Asians and Amerindians.[14] Our finding on the allele frequencies of ACP1 system (ACP1*A=0.250, ACP1*B=0.693, ACP1*C=0.057) corresponds with the previous data obtained in Turkey by Ates *et al*.[15] The figure of relatively high frequency of present ACP1*B

Table 1: The allele frequencies of red cell enzyme and serum proteins in Turkish population

System	Phenotype Observed	Frequency Expected	Allele Frequencies	
ACP1				
A	16	12.50	ACP1*A	0.250 ± 0.021
AB	60	69.25	ACP1*B	0.693 ± 0.023
B	102	95.91	ACP1*C	0.057 ± 0.011
AC	8	5.75		
BC	13	15.92		
C	1	0.66		
Total	200			
χ^2 = 4.16 *df*: 4 *P* > 0.05				
PGM1				
1	96	100.82	PGM1*1	0.710 ± 0.022
2-1	91	81.65	PGM1*2	0.288 ± 0.022
2	12	16.53	PGM1*6	0.002 ± 0.002
6-1	1	0.71		
6-2	0	0.29		
6	0	0.001		
Total	200			
χ^2 = 2.53 *df*: 2 *P* > 0.05				
GLO1				
1	38	27.82	GLO1*1	0.373 ± 0.024
2-1	73	78.62	GLO1*2	0.627 ± 0.024
2	89	93.56		
Total	200			
χ^2 = 1.99 *df*:2 *P* > 0.05				
ESD				
1	129	129.61	ESD*1	0.805 ± 0.019
2-1	64	62.79	ESD*2	0.195 ± 0.019
2	7	7.61		
Total	200			
χ^2 = 0.074 *df*:2 *P* > 0.05				
HP				
1	17	14.05	HP*1	0.265 ± 0.022
2-1	72	77.91	HP*2	0.735 ± 0.022
2	111	108.05		
Total	200			
χ^2 = 1.15 *df*:2 *P* > 0.05				
PI				
M1	160	157.54	PI*M1	0.887 ± 0.015
M1-M2	2	3.55	PI*M2	0.010 ± 0.004
M2	0	0.02	PI*M3	0.095 ± 0.014
M1-M3	30	33.72	PI*S	0.008 ± 0.004
M2-M3	2	0.39		
M3	3	1.80		
M1-S	3	2.66		
M2-S	0	0.03		
M3-S	0	0.29		
S	0	0.01		
Total	200			
χ^2 = 10.5 *df*: 1 *P* > 0.05				

allele and low frequency of ACP1*A allele resemble to those of Asians (ACP1*A=0.115-0.405, ACP1*B=0.546-0.885). On the other hand, the existence of ACP1*C allele indicates the genetic influence by Caucasoids.

PGM1

The distribution of PGM1*1 allele frequency is on the whole higher in Negroid and Mongoloid populations than in Caucasoid populations.[2] In the Caspian littoral populations, the frequency of PGM1*1 allele is rather low (0.585-0.705) than other Asian and European populations. The value observed for PGM1*1 allele in our Turkish population (0.710) is closer to the south and south east European populations such as Greeks, and Italians (0.68-0.71)[16-18] and fit well to the range for Turkey (0.567-0.759) reported in earlier studies.[19,20]

GLO1

Indians from North America, Chenese population and Negroes show a lower incidence of GLO1*1 (0.11-0.28). Japanese population shows much less heterogeneity for the GLO polymorphism. While the allele frequency of GLO1*1 has a mean value of about 0.305 in Asian populations, this value is about 0.44 in European populations as shown in Meighen *et al*'s study.[21] In the present study, frequency of GLO1*1 for Turkish population was found to be 0.373. This value is inbetween those of Asians and Europeans**.** It should also be pointed out that frequency of GLO1*1 allele is within the limits of previous Turkish studies,[19,22,23] which indicated the frequency to be between 0.363 and 0.428.

ESD

Cartwright *et al*.[24] and Ebeli-Struijk *et al*.[25] stated that ESD*1 allele frequencies of Asiatic Indians and Kuwait are intermediate between those of Europeans and Mongoloids and suggested that a genetic cline may exist across the Middle East. The present value of ESD*1 allele in Turkish population (0.805) is very close to that found for Kuwait population (0.803), and the same with Iranian Turkomans (0.805).[26] However, considerable variability in the frequency of ESD*1 allele was observed for Turkey in earlier studies, with figures ranging from 0.619 to 0.897.[19,27] The findings of the present study is in good agreement with those found in previous Turkish studies.

HP

Considering only the HP*1 allele frequencies of haptoglobin described in the literature, it may be said that the HP*1 frequency in our Turkish population (0.265) is quite similar to those of Asian populations (0.21-0.31) than Caucasians (0.38-0.42). Regarding the data of previous studies,[28-30] HP*1 allele frequency is between 0.265 and 0.275 in Turkey[28-30] and our results fit well in the lower end of this range.

PI

PI*M1 frequency in our sample (0.887) is somewhat higher than those reported for most of the European and Asian populations and resembles to those of northern European populations where the PI*M1 frequency is >80 (Sweden, Norway, Finland). The present PI*M2 frequency of 0.010 is quite lower than reported for most of Asian and European populations. The present value is similar to those of Asian populations where the range is 0.002-0.014 as shown in the study by Nevo *et al*.[31] PI*S allele occur only sporadically in other population samples than Caucasian populations.

The allele frequencies of 3 traits (ACP1, ESD and HP) are similar to those of Asians, whereas those of 2 traits (PGM1 and PI) are close to European populations. The allele frequency of GLO1 system is inbetween those of Asians and Europeans. In conclusion, the allele frequencies of Turkish population are closer to Asians than European populations. However, our Turkish population shows genetic influence of Europeans.

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