Detection of Some Blood Group (ABO, RH-D) and Serum Protein (HP, a1-AT, TF) Polymorphisms in Antakya Province, Turkey

ANTAKYA YÖRESİNDE KAN GRUBU (ABO, RH-D) ve SERUM PROTEİNİ (HP, a1-AT, TF) POLİMORFİZMİNİN İNCELENMESİ

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Summary

Objectives: In this study, we investigated ABO and RJi(D) blood groups polymorphisms and HP (Haptoglobin), a1-AT (al-antitrypsin-Protease Inhibitor-Pi) and TF (Transferrin) serum protein polymorphisms in Antakya province of Turkey.

Material and Methods: The ABO and Rh(D) blood group phenotypes of 2270 individuals who applied to the Blood Centre of Antakya Public Hospital for typing of their blood groups during the January-October 2001 were statistically evaluated and gene frequencies of blood groups were calculated. For the typing of serum proteins, about 5 ml venous blood sample taken from the 100 healthy and unrelated adults of both sexes. Plasma samples were obtained by centrifugation and stored at -20°C until phenotyping. While the HP phenotypes were determined by starch gel electrophoresis, a1-AT and TF phenotypes were detected by cellulose acetate paper electrophoresis.

Results: The calculated gene frequencies were as follows: A= 0.282, B=0.120, 0=0.598; D=0.719, d=0.281; HP* 1=0.275, HP*2=0.725; PI*M= 1,TF*C=1.

Conclusion: In conclusion, it may be said that the obtained gene frequencies of studied systems (ABO, Rh-D, HP, a1-AT, TF) were in agreement with those of overall Turkish data reported in earlier studies. Also the gene frequencies of two system (ABO and RH-D) were found to be inbetween those of Asians and Europeans. On the other hand, the gene frequencies of three traits (HP, a1-AT, TF) were similar or close to those of Asians.

Key Words: Blood groups, Serum proteins, Gene frequencies, polymorphic systems

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Özet

Amaç: Bu çalışmada, Antakya yöresinde ABO ve Rh(D) kan grupları ile HP (Haptoglobin), a1-AT (al-Antitripsin, Proteaz İnhibitör-PI) ve TF (Transferlin) serum proteinlerinin polimorfizmi araştırılmıştır.

Materyal ve Metod: 2270 bireyin ABO ve Rh(D) kan grubu fenotipleri, Antakya Devlet Hastanesi Kan Merkezi'nin veribankasından alınarak istatistiksel olarak değerlendirilmiş ve gen frekansları hesaplanmıştır. Serum proteini polimorfizmi ise; Antakya yöresinde yaşayan ve birbiri ile akrabalık ilişkisi bulunmayan 100 kişiden oluşan bir populasyon örneğinde araştırılmıştır. Bireylerin HP fenotipleri nişasta jel elektroforezi ile, a1-AT ve TF fenotipleri ise selüloz asetat kağıt elektroforezi ile saptanmıştır. Serum proteinlerinin gen frekansları, gen sayımı yöntemi ile hesaplanmıştır.

Bulgular: Çalışma sonucunda gen frekansları; A= 0.282, B=0.120, 0=0.598; D=0.719, d=0.281; HP*1=0.275, HP*2=0.725; PI*M= 1, TF*C=1 olarak hesaplanmıştır.

Sonuç: Çalışılan sistemlerin (ABO, Rh-D, HP, a1-AT, TF) gen frekanslarının Türkiye'de daha önce bulunan gen frekanslarına benzer olduğu söylenebilir. Ayrıca iki sistemin (ABO ve Rh-D) gen frekansları Asya ve Avrupa populasyonları arasında bir orta durum göstermektedir. Diğer taraftan HP, a1-AT ve TF gen frekansları Asya populasyonu ile örtüşmektedir.

Anahtar Kelimeler: Kan grupları, Serum proteinleri, Gen frekansları, Polimorfik sistemler

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The blood groups, red cell isoenzymes, serum proteins, hemoglobin variants, and the HLA system are the genetic markers in human blood used for the studying human genetic variation. The gene frequencies of these polymorphic systems may provide clues for genetic structure of a population and for the study of human evolution. Also these systems are usefull for establishing identity of an individual in forensic sciences (1-5).

Table 1. The phenotypes and gene frequencies of the ABO, Rh(D) blood groups and HP, a1-AT, TF serum protein systems in Antakya province

System	Observed number	Expected number	Phenofype frequencies (%)	Gene frequencies
ABO				
A	943	946.11	41.54	A=0.282±0.007
B	355	358.47	15.63	B=0.120±0.004
0	819	811.76	36.07	**O**=0.598±0.007
AB	153	153.63	6.74	
x^2= 0.1	df=l	p= 0.75		
Rh				
D	2090		92.07	D=0.719±0.006
d	180		7.92	d=0.281±0.006
HP				
l-l	8	7.56	8	HP*1=0.275±0.031
2-1	39	39.87	39	HP*2=0.725±0.031
2-2	53	52.56	53	
x^2= 0.03	*dM*	p=0.86		
a1-AT				
M-M	100		100	PI*M=1.00
TF				
C-C	100		100	TF*C=1.00

The aim of this study is the investigation of some blood group and serum protein polymorphisms in a population sample of residents in Antakya province to enlarge our knowledge on the distribution of gene frequencies in Turkey.

Materials and Methods

A population sample living around Antakya area was examined for the distribution of ABO and Rh(D) blood group and serum proteins namely, haptoglobin (HP), a1-antitrypsin (a1-AT, PI-Protease Inhibitor) and transferrin (TF) polymorphisms.

The ABO and Rh(D) blood group phenotypes of 2270 individuals who applied to the Blood Centre of Antakya Public Hospital for typing of their blood groups during the January-October 2001 were statistically evaluated and gene frequencies of blood groups were calculated (6).

For the typing of serum proteins, about 5 ml venous blood sample taken from the 100 healthy and unrelated adults of both sexes. Plasma samples were obtained by centrifugation and stored at -20°C until phenotyping. While the HP phenotypes were determined by starch gel electrophoresis (7), a1-AT and TF phenotypes were detected by cellulose acetate paper electrophoresis (4).

The gene frequencies were calculated by gene counting method and the possible divergence from Hardy-Weinberg equilibrium was examined using the x^2-test (8).

Results

The phenotypes and gene frequencies of the studied polymorphic systems are shown in Table 1.

As shown in the Table 1, ABO gene frequencies were estimated as A=0.282, B=0.120, 0=0.598. The most frequent phenotype was A

(41.50 %), and the second one was O (36.07 %) group. The system were found to be in good agreement with the Hardy-Weinberg equilibrium. The D gene frequency of Rh blood group was calculated as 0.719 (Table 1). The Rh positive (+) phenotype was shown in proportion of 92.07%.

In the serum protein system HP, all common phenotypes were detected and HP*1 and HP*2 alleles were observed with the frequencies of 0.275 and 0.725 respectively (Table 1). The HP system was in good agreement with the Hardy-Weinberg equilibrium. The a1-AT serum protein system found to be monomorphic and only the PI M-M phenotype was observed. Also the serum protein TF did not show variation and all phenotypes were TFC-C (Table 1).

Discussions

Blood Groups (ABO, Rh-D)

ABO: There are several studies on the distribution of ABO gene frequencies in Turkey. In a previous study, Aytaç (9) described the gene frequency variations in eight region of Turkey and stated the gene frequencies to be 0.2842 for A, 0.1252 for B and 0.5933 for O in Turkey. In another study, Togan and Ergiiven (10) investigated the ABO gene frequencies in five region of Turkey and reported the frequency distribution of A, B, O genes to be as 0.3218, 0.0951, 0.5831 respectively for the overall Turkish data. Moreover Önde and Kence (11) determined the gene frequencies of ABO blood group in 67 province of Turkey. These studies indicated the A gene frequency ranges from 0.2014 to 0.3873, B gene from 0.0829 to 0.1656 and the O gene from 0.5222 to 0.6840 in Turkey. In this population sample residing in Antakya area, ABO gene frequencies were calculated as follows: A=0.282, B=0.120, 0=0.598 (Table 1). Tarskaia et al. (12) reported that the ABO gene frequencies were concluded as A=0.221, B=0.223 and 0=0.588 in Yakuts. The present values of ABO genes was within that range and were quite similar to the values (A= 0.271, B= 0.129, 0= 0.600) found by Önde and Kence (11) in Antakya area and by Tarskaia et al. (12) in Yakuts. Moreover, the present ABO gene frequency distribution in Antakya province was inbetween those of Europeans and Asians as indicated in the study of Togan and Ergiiven (10).

Rh (D): Earlier studies showed the D gene frequency ranges between 0.535-0.737 in populations living in Turkey (9-11). Togan and Ergiiven (10) indicated the considerable variability of D gene frequency. Önde and Kence (11) stated the sources of this heterogeneity were Eastern Black Sea and Southern Anatolia. In this study, The D gene frequency was calculated as 0.719 (Table 1). The present D gene frequency fitted well in the upper end of that range in Turkey. Also it was similar to the value (0.723) of previous study in Antakya area (11) and was between to those of Asian and European populations.

Serum Protein Systems (HP, a1-AT, TF)

HP: In this population sample no rare electrophoretic HP variants were detected. The HP*1 gene frequency was estimated as 0.275 (Table 1). The similar results were obtained in Yakuts (12). In a previous study, Erdem and Aksoy (13) reported the HP*1 gene frequency to be 0.265 in Turkey. Also Dönbak et al. (14) found the same HP*1 frequency (0.265) in Çukurova region of Turkey. On the other hand, Alper et al (15) obtained a higher value of HP*1 frequency (0.290) in Çukurova region than the one reported in the latter study. The present value of 0.275 was not different from the overall Turkish data. Moreover, the present HP*1 frequency was found to be within the reported range of Asian population (0.21-0.31) and it was similar to those of some populations living in Iran (i.e.. 0.270 in Bandaris, 0.280 in Turkomans)(16). Hamad and Awadallah (17) reported that the HP polymorphisim varied in different age groups in Jordanians like HP*2-2 gradually decreased while HP*2-1 gradually increased due to the age groups. In addition, Kasvosve et al. (18) reported that HP reference values were found to be HP phenotype-dependent in Black-Zimbabwean population that the highest values were found in HP*1-1.

a1-AT: In this study, only the PI*M allele was observed. The PI*Z and the PI*S rare alleles were

not detected and PI system found to be monomorphic. This observation was agreed with that of previous study made by Kökdener (19). PI*M allele is the most common allele of the system in all the populations. The same results were found by Tarskaia et al (12). The rare alleles, PI*Z and PI*S observed in polymorphic frequencies in Caucasians. It was reported that there is a geographical distribution on S and Z gene frequencies in Europe (20, 21). The frequencies of PI*Z and PI*S allele found to be 0.02-0.06 and 0.02-0.03 in Europe respectively. These alleles are seen rarely in other populations than in Caucasians (22,23). In this population sample, PI*Z and PI*S alleles were not observed. It may be due to small sample size.

TF: No TF variation was found in this population sample consists of 100 individuals. All specimens showed the TF C-C phenotype. The rare alleles of the system, TF*B and TF*D were not detected. Also Dinçol et al (24) found the system to be monomorphic in Turkish population and they observed only the TF*C gene. The frequency of TF*C gene was found to be higher than 0.95 in all human populations. While TF*B allele was not observed in the Asian population, the TF*D allele frequency have been found relatively higher than Europeans as shown in the study of Ohkura et al (25). In Yakuts, TF*C allele was found between 0.7 to 1.0 (12). In a previous study, Kalfoğlu and Atasoy (26) observed the TF*D allele with a frequency of 0.001 in Turkey. They investigated a population sample consist of 500 individuals and reported the TF*C allele frequency to be as 0.999 in Turkey. Also TF*D gene was observed in Eti-Turks living Southern part of Turkey by Dinçol et al (27). Althought Eti-Turks were residing in Antakya province, in this study, TF*D gene was not observed mainly due to small sample size.

In conclusion, it may be said that the obtained gene frequencies of studied systems (ABO, Rh-D, HP, a1-AT, TF) were in agreement with those of overall Turkish data reported in earlier studies. Also the gene frequencies of two system (ABO and RH-D) were found to be inbetween those of Asians and Europeans. On the other hand, the gene frequencies of three traits (HP, a1-AT, TF) were similar or close to those of Asians.

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