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Phenotypic and Genetic Frequency of the Erythrocyte Enzyme Systems ADA and AK in Bulgarians Living in the South Central Region of Bulgaria

S. Baltova

Department of Human Anatomy and Physiology, Faculty of Biology, University of Plovdiv, Plovdiv

For the study of ADA and AK we made parallel investigations. We took blood samples from 2368 individuals with Bulgarian origin, males and females, at the age from 18 to 45, clinically healthy and without family relationships among them. The phenotypes of ADA and AK were determined through horizontal electrophoresis of haemolysates on starch gel, according to the method of Radam and Strauch [1968]. The most frequently phenotypes are ADA 1-1 — 85.35% and AK 1-1 — 93.54% followed by the heterozygous. The genetic frequencies of the alleles from ADA and AK are as follows: ADA^1 =0.9240, ADA^2 =0.0760; AK^1 =0.9675, AK^2 = 0.0325.

Key words: polymorphism, ADA, AK, phenotype, gene frequency.

Introduction

We still do not have sufficient information for the expression of the genetically determined polymorphism of the enzymes of the Bulgarian population.

There are population and genetic studies of the erythrocyte enzyme systems adenosine-desaminase (ADA) and adenylat-kynase (AK) for certain cities — Sofia and Plovdiv but there is missing data about other regions in Bulgaria. [1, 7, 8].

The aim of the present study is to determine the phenotypic distribution and the genetic frequency of ADA and AK for the clarification of the genetic status of the Bulgarian population in the South central region of Bulgaria and to make a comparison with other populations.

Material and Methods

For the study of ADA and AK we made parallel investigations. We took blood samples from 2368 individuals with Bulgarian origin, living in the South Central region of Bulgaria, males and females, at the age from 18 to 45, clinically healthy and without family relationships among them. The distribution of the investigated indi-

viduals in sub-populations is as follows: the region of Plovdiv — 1508 individuals, the region of Stara Zagora — 450 and the region of Haskovo — 410.

The phenotypes of ADA and AK were determined through horizontal electrophoresis of haemolysates on starch gel, according to the method of Radam G., Strauch [5, 6].

Results and Discussion

In all the investigations we found out only the classic types of ADA and AK (Table 1). The phenotype 1-1 of ADA and AK has the greatest distribution in the studied quota — 2368 individuals. It is found respectively in 2021 individuals (85.35%) for ADA and in 2215 individuals (93.54%) for AK. The phenotypic distribution in the different sub-populations shows that phenotype 1-1 for both enzymes decrease its frequency in direction — regions of Plovdiv, Stara Zagora and Haskovo.

In Table 2 we represent the genetic frequencies of the enzymes ADA and AK. The genes from locus ADA and AK that determine homozygous phenotypes 1-1 and 2-2 are characterized from alleles with frequency for the population of the whole studied region respectively: $ADA^1 = 0.9240$, $ADA^2 = 0.0760$ and $AK^1 = 0.9675$, $AK^2 = 0.0325$. For the different sub-populations we observe increase of the genetic frequency of the allele ADA^1 and AK^1 in direction - regions of Plovdiv, Stara Zagora and Haskovo.

Table 1. The observed and expected phenotype frequencies in ADA and AK enzyme system in the samples studied

Region	Parameters	ADA			AK		
		phenotype 1-1	phenotype 2-1	phenotype 2-2	phenotype 1-1	phenotype 2-1	phenotype 2-2
Plovdiv	Observ.	1306	196	6	1419	88	1
	%	86.60	13.00	0.40	94.10	5.84	0.06
	Expect.	1307.08	193.74	7.18	1419.46	87.20	1.34
Stara Zagora	Observ.	376	70	4	419	31	0
	%	83.55	15.56	0.89	93.11	6.89	—
	Expect.	375.36	71.26	3.38	419.57	29.89	0.54
Haskovo	Observ.	339	68	3	377	33	0
	%	82.68	16.59	0.73	91.95	8.05	—
	Expect	339.37	67.29	3.34	377.70	31.64	0.66
Total region	Observ.	2021	334	13	2215	152	1
	%	85.35	14.10	0.55	93.54	6.42	0.04
	Expect.	2021.74	332.58	13.68	2216.58	148.92	2.50

Table 2. The gene frequencies in ADA and AK enzyme systems in samples studied

Alleles	Plovdiv $n = 1508$	Stara Zagora $n = 450$	Haskovo $n = 410$	Total region $n = 2368$
ADA^1	0.9310	0.9133	0.9098	0.9240
ADA^2	0.0690	0.0867	0.0902	0.0760
$\chi 2$	0.0264	0.0223	0.0075	0.0061
	$0.05<P<0.20$	$0.5<P<0.80$	$0.99<P<0$	$0.05<P<0.20$
AK^1	0.9702	0.9656	0.9598	0.9675
AK^2	0.0298	0.0344	0.0402	0.0325
$\chi 2$	0.0073	0.0412	0.5400	0.5820
	$0.05<P<0.20$	$0.05<P<0.20$	$0.20<P<0.50$	$0.01<P<0.05$

Table 3. Distribution of gene frequencies of ADA and AK by others populations

Population	Locus ADA			Locus AK			Authors
	No	ADA^1	ADA^2	No	AK^1	AK^2	
Sofia	138	0.8623	0.13760,	138	0.9637	0.03620,	Ananthakrishnan, et al. (1972)
	402	0.9440	0560	310	0.9710	0390	Попвасилев, И. (1980)
	2694	0.9311	0.0689	—	—	—	Ненков, Н. (1981)
Plovdiv	1508	0.9310	0.0690	1508	0.9702	0.0298	Ours studies
	116	0.9138	0.0862	—	—	—	Ненков, Н. (1981)
St. Zagora	450	0.9133	0.0867	450	0.9656	0.0344	Ours studies
Haskovo	410	0.9098	0.0902	410	0.9598	0.0402	Ours studies
Kyustendil	100	0.9500	0.0500	—	—	—	Ненков, Н. (1981)
Gipsy	171	0.8655	0.1345	—	—	—	Ненков, Н. (1981)
Germany	2925	0.9460	0.0540	1067	0.9610	0.0390	Schel, H-G. (2001)
Hungary	654	0.9450	0.0550	654	0.9680	0.0320	Pap, M. (2000)
Europe	—	0.9400	0.0600	—	0.9500	0.0500	Lewontin, R.C. (1974)
Africa	—	0.9700	0.0300	—	1.0000	0.0000	Lewontin, R.C. (1974)
India,Pakistan	—	0.8600	0.1400	—	0.8700	0.1300	Lewontin, R.C. (1974)
Andi-redskin	—	1.0000	0.0000	—	1.0000	0.0000	Lewontin, R.C. (1974)

We used the criterion of Pierson and we compared the distribution among the observed and the expected values and we found out that the difference is not significant — $P>0.05$, $\chi^2=0.402$ for ADA and $\chi^2=0.9648$ for AK. The correlation between the observed and the expected values is good and according to the law of Hardy-Weinberg shows that the studied population is in genetic balance concerning the enzymes ADA and AK.

We compared the results of the present study with these of other populations made by different authors according to literary data (Table 3).

The values of the frequency of the allele ADA^2 is zero in Negroes, South-American Indians and from 0.11-0.16 for South Asia. For Europe the ADA^2 frequency in the different populations vary in narrow borders (0.05-0.09) and it has certain tendency to increase from North to South [3, 6, 8].

In Europe we have predominantly the gene AK^1. The frequency of the alleles AK^1 and AK^2 in the European populations has certain stability — AK^1 - 95-97% and AK^2 — 2.9-4.5%. In the Negroid and Mongoloid populations the frequency of the allele AK^2 is considerably lower than in the Europoid populations.

Conclusions

1. The most frequently phenotypes are ADA 1-1 — 85.35% and AK 1-1 — 93.54% followed by the heterozygous. The genetic frequencies of the alleles from ADA and AK are as follows: $ADA^1=0.9240$, $ADA^2=0.0760$; $AK^1=0.9675$, $AK^2=0.0325$.

2. The frequency of the iso-enzyme variations in the studied Bulgarian population does not differ substantially from the European populations. We observe a tendency of west-east and north-south geographic distribution.

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