

Red-Cell Adenylate Kinase 1 Polymorphism in Turkey

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TÜRKİYE'DE ALYUVAR ADENİLAT KİNAZ 1 POLİMORFİZMİ

Özet

Biyolojik babanın saptanması ve vücut sıvılarının individualizasyonu amacıyla Türkiye'de alyuvar adenilat kinaz 1 (AK1) enzim sisteminin fenotip dağılımı ve gen frekansları araştırıldı. Bu amaçla akrabalık ilişkisi bulunmayan 359 kişi incelendi. Adenilat kinaz 1 sistemi için elde edilen alel frekansları şöyledir: $AK1*1 = 0.954 \pm 0.008$ ve $AK1*2 = 0.046 \pm 0.008$ ($\chi^2 = 0.0731$; D.F. = 1; $0.7 < p < 0.8$).

Summary

A sample of 359 unrelated individuals from Turkey was studied for the red-cell adenylate kinase 1 (AK1) polymorphism. The following allele frequencies were observed: for $AK1*1 = 0.954 \pm 0.008$ and $AK1*2 = 0.046 \pm 0.008$ ($\chi^2 = 0.0731$; D.F. = 1; $0.7 < p < 0.8$).

Key words: Red-cell adenylate kinase - Polymorphism - Turkey

INTRODUCTION

Red blood cell adenylate kinase 1 (AK1) polymorphism has been used successfully for blood and bloodstain individualization in criminalistic laboratories (1-3). Although there are some rare phenotypes reported, generally the three distinct phenotypes of AK1 are AK 1-1, AK 2-1 and AK 2-2, directed by two common autosomal alleles $AK*1$ and $AK*2$ (4,6). Due to the fact that it is a very stable enzyme in various conditions, conventional electrophoresis methods give satisfactory results and there are several methods proposed by different studies (3,7,8). There is a good amount of information about AK allele frequencies in European, American and some Asiatic nations available, however there has been very limited work done on the genetic polymorphism of red-cell AK1 in Turkey (9,10).

Adli Tıp Derg., 6, 69 - 71 (1990)

The purpose of the following study was to estimate gene frequencies of the enzyme in order to be used in criminalistic cases and paternity disputes. Part of this work was published elsewhere (11,12).

MATERIAL and METHODS

Blood samples of 359 unrelated individuals were examined for AK1 phenotypes.

Hemolysates were prepared by first centrifuging the fresh blood samples that were taken with sodium citrate as anticoagulant and separating the plasma. The red blood cells were diluted to a ratio of 1:2.5 with distilled water, frozen and then thawed twice to achieve complete lysis and stored in -20°C till the date of investigation which was within a week after the collection of the samples.

AK1 was phenotyped by conventional cellulose acetate electrophoresis using a 0.014 M phosphate buffer, pH 6.25 both as tank and membrane buffers. Electrophoresis was performed at a constant voltage of 300 V for 45 min, at room temperature. The staining was carried according to *Grunbaum* as modified by *Abacı* (11). The reaction mixture consisted of 38 mg adenosine-5-diphosphate, 190 mg dextrose, 12 mg nicotinamide adenine dinucleotide phosphate (NADP), 8.75 units glucose-6-phosphate dehydrogenase, 7.0 units hexokinase, 2 mg 3-(4,5-dimethyl thiazolyl-2)-2,5 diphenyl tetrazolium bromide (MTT) tetrazolium, and 2 mg phenazine methosulphate (PMS) dissolved in 15 ml of 0.014 M phosphate buffer (pH 6.25). 200 mg agar were dissolved in 10 ml of the same buffer and the two solutions were mixed when the latter was 55°C. The above reaction mixture was then poured to square petri dishes. After it solidified it was incubated with the cellulose acetate membrane of the electrophoretic run for 10 minutes. The adenylate kinase isoenzymes are visible as blue bands on a clear background.

Gene frequencies were calculated by gene counting and departures from the Hardy-Weinberg equilibrium by χ^2 .

Table I. Phenotype and gene frequencies of red-cell adenylate kinase 1 in Turkey

<i>Genotypes</i>	<i>Observed</i>	<i>Expected</i>	<i>Allele Frequencies</i>
AK 1-1	326	326.73	$AK1*1 = 0.954 \pm 0.008$
AK 2-1	33	31.50	$AK1*2 = 0.046 \pm 0.008$
AK 2-2	0	0	

$\chi^2 = 0.0731$; D.F. = 1 ; $0.7 < p < 0.8$

RESULTS and DISCUSSION

The gene frequency distribution of the enzyme found to be as follows: $AKI*1 = 0.954 \pm 0.008$ and $AKI*2 = 0.046 \pm 0.008$ (Table I). Observed and expected values confirmed with *Hardy-Weinberg* equilibrium ($\chi^2 = 0.0731$) The phenotype distribution of 359 unrelated adults from all parts of Turkey is comparable with that found in other European populations. These results are also consistent with the AK1 gene frequencies presented by *Hummel et al* (9), which is concerned about gene frequencies in Turkish people living in Germany ($AKI*1 = 0.9580$, $AKI*2 = 0.420$, $n = 274$) and *Menevşe* and *Ülküer* ($AKI*1 = 0.969$, $AKI*2 = 0.031$, $n = 116$) (10).

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