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Original scientific paper

Genetic Studies in South Balkan Populations

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A B S T R A C T

Within a study of the genetics of Balkan populations, four DNA-STR systems and 19 classical markers were examined in seven samples: Romanians (two groups), Albanians, Greeks and Aromuns (three groups). The results for the DNA-STR systems have been compared with data from the literature. The results show four clear separated groups: sub-Saharan black populations, North-African, Japanese and European populations. The large Balkan populations, except the Greek sample, are genetically more homogenous than the Aromun populations. A second Neighbor-joining tree based on all 23 analyzed systems, show a particular trend of the Aromun groups, which indicates a particular genetic structure.

Key words: *population genetics, Balkan, Aromuns, DNA-STR marker D21S11, FGA, TH01, VWA, blood groups, serum protein polymorphisms, red cell enzyme polymorphisms*

Introduction

The present study deals principally with the genetic relationships of Balkan populations. Up to now there is little known on the distribution of genetic markers in South-Eastern Europe.

An analysis of 23 systems has been done in seven populations from the Balkans, including Romanians, Albanians, Greeks and three Aromun populations from Albania, Romania and the Republic

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of Macedonia. Four DNA-STR systems of seventeen European, African and Asian samples, published by other authors, are included[1].

At present, the Aromuns are geographically distributed over all Balkan countries[2]. Most of them speak a dialect derived from classical Latin[3]. Their origin is still unclear and they are considered as relict groups of a formerly widespread and continuous large population. The Aromuns can be divided into four larger groups based on ethnic and lingual differences:

1. The *»classical Aromuns«*. Their settlement areas are spread all over the Balkans.
2. The *Megloromuns*, that have been drastically reduced in size. They live merely in two small areas near the Greek-Macedonian border and in Romania (Dobruja province).
3. The *Istroromuns*, whose settlements lies far from other Aromuns in Istria (Croatia).
4. The *Sarakatsans,* who are spread all over the Southern Balkan Peninsula.

The *»classical Aromuns«* represent the largest group. Based on their geographical origin and distribution, they can be divided into six groups: *Pindonians, Gramostians*, *Moskopolians, Verians, Fraseriots* and *Musequiars*.

Material and Methods

Four ethnic groups were analyzed in this study: three *»classical Aromun«* populations, two samples from Romania, one Albanian population and one from Greece (Figure 1). The samples of Aromun populations come from Romania, village Kogalniceanu (Fraseriots, 99 individuals), from Albania, village Andon Poci (Pindonians, 100 individuals) and from the Republic of Macedonia, region Stip (Gramostians, 108 individuals). The blood samples of Albanian population (99 individuals) were collected in the capital Tirana. The Romanian samples were collected in the cities Constanta (146 individuals) and Ploiesti (126 individuals). The Greek sample (108 individuals) come from the North-Eastern region of Greece (Thracia).

Fig. 1. Geographical distribution of the population samples studied: Aromuns: 1 = Pindonians (Albania, Andon Poci), 2 = Gramostians (Macedonia, Stip), 3 = Fraseriots (Romania, Kogalniceanu), 4 = Albanians (Tirana), 5 = Greeks (Thracia), 6 = Romanians (Constanta), 7 = Romanians (South, Ploiesti).

In all samples four DNA-STR systems (D21S11, FGA, TH01, VWA), four blood groups (ABO, RH, P1, Kk), seven serum protein polymorphisms (GC, HPA subtypes, CP, TF, AMY2, BF, C3) and eight polymorphisms of erythrocyte enzymes (PGM1, PGM3, GPT, GOT2, GLO, ESD, ACP, UMPK) were tested according to standard procedures.

The allele frequencies were calculated by direct gene counting, except ABO[4], RH[5], P1[5] and GPT[6]. Allele frequencies of the four DNA-STR systems and for the seven serum proteins had been published

elsewhere[7,8]. The allele frequencies for the blood groups and erythrocyte enzymes for the South-Romanians from Ploiesti, for the Greeks from Thracia and for the Aromuns from Stip (Republic of Macedonia) are listed in Table 1. Data for all other populations (Romanians from Constanta, Albanians, and Aromuns from Romania (Fraseriots) and Albania (Pindonians) had been published previously[9].

To show the relationships between all populations studied Neighbor-joining trees using Reynolds genetic distances were depicted and a bootstrap analysis was done with 1,000 replicates.

Results and Discussion

The first tree (Figure 2) shows the genetic relationships between all popula-

TABLE 1
ALLELE AND HAPLOTYPE FREQUENCIES OF THE THREE UNPUBLISHED SAMPLES (HWE: TEST OF HARDY-WEINBERG EQUILIBRIUM, DF = 0: NO DEGREES OF FREEDOM FOR TESTING)

	Greeks (Thracia)	Romanians, South (Ploiesti)	Aromuns, Gramostians (Macedonia, Stip)
A1	0.2731	0.2170	0.3054
A2	0.0201	0.0410	0.0201
B	0.0819	0.1082	0.1124
O	0.6249	0.6339	0.5621
N	141	126	108
HWE	df = 0	χ^2 = 1.9336, df = 1 10 < p < 20	df = 0
cde	0.3683	0.3714	0.2781
cDe	0.0435	0.0333	0.0136
Cde	0.0135	0.0223	0.0140
cdE	0.0120	–	0.0098
CDe	0.4339	0.4380	0.4812
cDE	0.1057	0.1230	0.1754
$C^{W}De$	0.0098	0.0119	0.0139
$CD^{U}e$	0.0135	–	0.0140
N	102	126	108
HWE	df = 0	χ^2 = 0.9474, df = 1 30 < p < 50	df = 0
P1+	0.3467	0.5040	0.5385
P1-	0.6533	0.4960	0.4615
N	164	126	108
K	0.0490	0.0357	0.0187
k	0.9510	0.9643	0.9813
N	102	126	107
HWE	df = 0	df = 0	df = 0
PGM1*1	0.5833	0.6429	0.5926
PGM1*2	0.2407	0.2381	0.2454
PGM1*3	0.1204	0.0873	0.0741
PGM1*4	0.0556	0.0318	0.0880
N	108	126	108
HWE	χ^2 = 1.4357, df = 2 80 < p < 90	χ^2 = 0.9065, df = 2 50 < p < 70	χ^2 = 0.9532, df = 2 50 < p < 70

CONTIN. OF TABLE 1

	Greeks (Thracia)	Romanians, South (Ploiesti)	Aromuns, Gramostians (Macedonia, Stip)
PGM3*1	0.7083	0.7976	0.7824
PGM3*2	0.2917	0.1984	0.2176
PGM3*VAR	–	0.0040	–
N	108	126	108
HWE	$\chi^2 = 3.1519$, df = 1	df = 0	$\chi^2 = 2.6662$, df = 1
	$5 < p < 10$		$10 < p < 20$
GPT*1	0.4904	0.5186	0.5303
GPT*1M	0.0189	0.0171	0.0206
GPT*2	0.4907	0.4643	0.4491
N	108	126	108
HWE	df = 0	df = 0	df = 0
GOT2*1	1.0000	0.9921	0.9861
GOT2*2	–	0.0079	0.0139
N	108	126	108
HWE	–	df = 0	df = 0
GLO*1	0.4167	0.3810	0.3287
GLO*2	0.5833	0.6191	0.6713
N	108	126	108
HWE	$\chi^2 = 0.2448$, df = 1	$\chi^2 = 0.0118$, df = 1	$\chi^2 = 0.0854$, df = 1
	$50 < p < 70$	$80 < p < 90$	$70 < p < 80$
ESD*1	0.8750	0.9087	0.9583
ESD*2	0.1111	0.0913	0.0417
ESD*5	0.0139	–	–
N	108	126	108
HWE	df = 0	df = 0	df = 0
ACP*A	0.2685	0.3254	0.3796
ACP*B	0.6435	0.6032	0.5833
ACP*C	0.0880	0.0714	0.0370
N	108	126	108
HWE	$\chi^2 = 1.9659$, df = 2	$\chi^2 = 4.5546$, df = 2	$\chi^2 = 2.4139$, df = 1
	$30 < p < 50$	$10 < p < 20$	$10 < p < 20$
UMPK*1	0.9861	0.9484	0.9722
UMPK*2	0.0139	0.0516	0.0278
N	108	126	108
HWE	df = 0	df = 0	df = 0

tions based on the four studied DNA-STR systems. The tree divides the populations into four groups: sub-Saharan black populations, North-African, Japanese and European populations. The black populations are clearly separated from the other populations with a very strong bootstrap support (95.2%). The North-African populations show a closer relationship to the European populations than to the sub -Saharan populations. The seven populations analyzed in this study belong clearly to the European cluster. The separation of the Greek sample from North-Eastern Greece is based mainly on the high frequencies of the DNA-STR alleles D21S

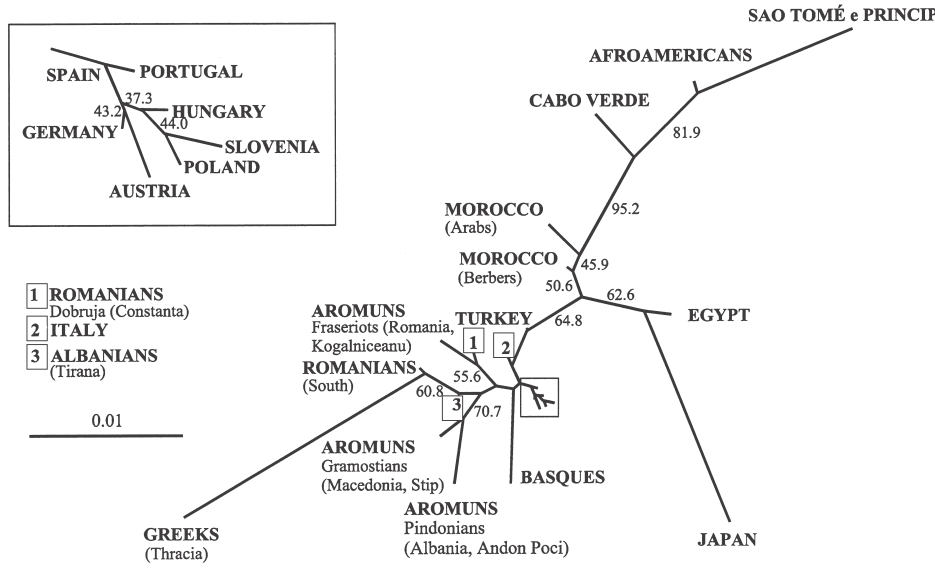

Fig. 2. Neighbor-joining tree of all populations studied (DNA-STR systems D21S11, FGA, TH01, VWA). The bootstrap supports of the branches are given beside the branches.

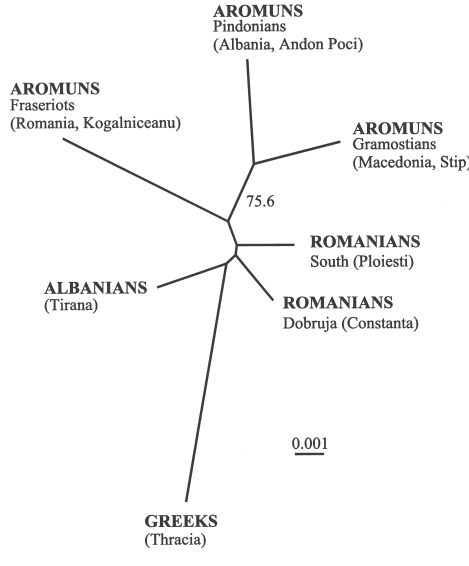

Fig. 3. Neighbor-joining tree of the Balkan populations studied (four DNA-STRs and 19 classical markers).

11*29 and FGA*22,2[7]. The Aromuns, like the Basques, show also a bigger genetical distance to other European populations and to each other. But generally, the European populations are genetically more homogeneous.

The other tree (Figure 3) shows the genetic relationships between Balkan populations. The genetical distances were calculated from the gene frequencies of the four DNA-STR systems and the 19 classical markers. The Aromun populations never show a close genetic relationship to each other. The genetic distances between them are always higher than between the large European populations. The large Balkan populations, except the Greek sample from Thracia, display also a higher genetic homogeneity between each other than the Aromun populations.

The results suggest a special position of the Aromuns with regard to their ge-

netics. This special position could already be shown in previous studies[9,10]. A more detailed understanding is expected from the extension of the present study based on additional samples and additional genetic markers (Alu-insertions, Y-chromosomal markers and mtDNA).

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GENETIČKE STUDIJE POPULACIJA JUŽNOG BALKANA

SAŽETAK

U sklopu studije genetike stanovništva Balkana, istražena su četiri DNK-STR sustava i 19 klasična markera u 7 populacijskih uzoraka: Rumunja (dva uzorka), Albanaca, Grka i Aromunja (3 uzorka). Rezultati DNK-STR sustava uspoređeni su s podacima iz literature. Rezultati su pokazali četiri jasno odvojene populacijske skupine: subsaharska, sjeverno-afrička, japanska i europska. Velike Balkanske populacije, osim uzorka iz Grčke, genetski su homogenije od Aromunjskih populacija. Drugo »neighbor-joining« stablo, temeljeno na sva 23 analizirana sustava, pokazalo je zaseban trend Aromunjske skupine, što ukazuje na posebnu genetsku strukturu ove populacije.