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Research Article

RELATIONSHIP BETWEEN EPIDERMAL RIDGE PATTERNS IN THREE COHORTS OF BULGARIAN POPULATION: A PILOT STUDY

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Summary: One of the main problems of population genetics is to precisely describe the genetic difference between two closely related species as compared to the genetic difference between two populations of the same species. Traditional population genetic analyses include the distribution of allele frequencies between and within populations. Using these frequencies, we can estimate the genetic characteristics of the population.

The fingerprints model (epidermal ridges) has become one of the most important indicators in determining genetically related groups in multiethnic populations. The aim of our pilot study was to carry out a dermatoglyphic analysis of two generations from 1947 (Markov, 1947) and 2012 (Angelova, 2012), respectively.

The two generations were situated in the same region of Northern Bulgaria. They included people aged from 16 to 60. The population from 2012 included 353 individuals from both genders. We categorized the population into three cohorts/communities - Bulgarian Christians, Bulgarian Muslims and Bulgarian Turks. The population from 1947 included 336 individuals and was used as control group for comparison of differences in fingerprint patterns. The parameters observed were loops, arches and whorls and were counted from the fingers of both hands.

The relationship between the fingertip patterns of the two populations showed significant variations. The comparison between the two generations of the Bulgarian Christians showed significant variations in all three dermatoglyphic patterns. In Bulgarian Muslims cohorts significant variations were found in the loops and whorls. No significant variation was detected in arches patterns in Bulgarian Muslims, while loops and whorls showed statistically significant variation.

Keywords: Dermatoglyphics, fingertip patterns, Bulgarian population, frequencies.

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INTRODUCTION

Evolution is the process by which populations change over generations. Genetic variations underlie these changes. The genetic structure of a population can evolve from generation to generation. Different characteristics tend to exist within any given population because of mutations, genetic recombinations and other sources of genetic variation. In addition, allele frequencies in populations also change due to natural selection (including sexual selection), and the genes flow from one population to another (migrations) and from the genetic drift.

In the last decade, the research of fingerprint models has become one of the most important indicators in identifying genetically related groups in multiethnic populations. Studies show differences in pattern among ethnic groups within one population (Damarchi et al., 1998; Adetona et al., 2008; Kobyliansky et al., 1988, 2004; Karmakar et al., 2009a, 2009b, 2009c).

It is well established that dermatoglyphics are genetically determined, but to date, few studies have given attention to the inheritance pattern of dermatoglyphics. Thus, the genetic nature of dermatoglyphics remains still unclear.

Dermatoglyphic patterns are genetically determined and follow a polygenic pattern of inheritance (Froehlich J, 1976). There are important questions concerning their inheritance like sexual dimorphism (Karmakar et al., 2002, 2008, 2009c; Scheil et al., 2005), their ontogeny (Babler,1991), health conditions (Buković et al., 1999; Burute et al., 2013; Kobyliansky et al.,1997, 1999, 2004, 2005; Rogucka and Hauser, 1998), chromosome aberrations (Jantz and Hunt, 1986; Bat-Miriam Katznelson et al., 1999), and asymetry (Buchwald, 2002, 2015; Andreenko and Baltova, 2015). The dermatoglyphic patterns are more often a result of genetic influence than of the physical environment. Epidermal ridges lay down between the tenth and eighteenth week of gestation and remain unchanged throughout the whole period of life except for increasing in size (Lacroix and Wolff-Quenot, 1984; Milić and Pavićević, 2000;Stevenson and West, 2001;Wertheim, 2011; Godwin et al., 2016). Ridge patterns develop first on finger pads, then on palms, and finally on feet.

The crucial period in differentiation is the third embryonic month, and if different artefacts interfere in the ridge formation, they will affect their normal differentiation. Damages to the ridges and deviation from their normal formation can be due to genetic or prenatal factors (Meier et al., 1987; Sorenson et al., 1990; Andreenko and Baltova, 2015). Despite the key role of the genes responsible for dermatoglyphic expression, some aspects of dermatoglyphic variation can be also due to environmental factors such as radiation, medications, hormones and epigenetic changes (Meier, 1981; Milić and Pavićević, 2000).

The entire human body is covered with skin, which is the largest and most important organ. The skin on the ventral sides of the hands and feet is corrugated with ridges and different configurations (Kumbani, 2007; Robert E., 2005; Talwar et al., 2017; Kumar et al., 2017). All studies of these dermal models are classified under the term dermatoglyphics (Katznelson and

Goldman, 1982). Harold Cummins (1926) coined the term dermatoglyphics, which means "cuts on the skin". Sir Francis Galton formulated the complete system of fingerprint classification rules in 1892.

The ridge patterns on the distal phalanges of the fingertips are divided into three groups: arches, loops, and whorls (Fig. 2):

i) Arches (A) are the simplest pattern found on fingertips. It is formed by succession of more or less parallel ridges, which traverse the pattern area and form a curve that is concave proximally. The arch pattern is subdivided into two types: plain arch and tented arch.

ii) Loops (L) are the most common pattern on the fingertip. A series of ridges enter the pattern area on one side of the digit, recurve abruptly, and leave the pattern area on the same side. The loop pattern is subdivided into two types: ulnar loop and radial loop.

iii) Whorls (W) are any ridge configuration with two or more tri-radii. One tri-radius is on radial and the other one - on the ulnar side of the pattern. Subtypes of whorl patterns include simple/ concentric whorl, spiral whorl, central pocket whorl, lateral pocket/twinned loop pattern and accidental patterns.

In the last few decades, the medical interest in epidermal ridges has grown considerably because it was found that many patients with chromosome aberrations had unusual ridge patterns (Hoefnagel et al., 1963; Suzumori, 1980; Kobyliansky et al., 1997; Jeewandeep et al., 2013). Man can change, his behavior can change, thoughts can change, but the model of dermatoglyphics will remain the same until death. The study of admixed populations can contribute to understanding the genetic traits of dermatoglyphic patterns (Cheng et al., 2009).

If we want to determine how allele and genotype frequencies change in a population in time and in generations, we use the Hardy-Weinberg law. This law states that allele and genotype frequencies in a population will remain constant from generation to generation in the absence of other evolutionary influences. However, the influences like genetic drift, natural selection, mate choice, gene flow, assortative mating, mutation, meiotic drive, genetic hitchhiking, population bottleneck, founder effect and inbreeding always exist, and without them evolution would not be possible. Therefore, the Hardy-Weinberg law is the starting point for much of the theory of population genetics.

In the present study, a sample of individuals was used to analyze the relationship between dermatoglyphic patterns of three groups of the Bulgarian population - Bulgarian Christians, Bulgarian Muslims and Bulgarian Turks of two generations - from 1947 (Markov) and 2012 (Angelova). We used the three general fingerprint classification types: arches (A), loops (L) and whorls (W) of the left and right hands without consideration of their variations (Aigbogun E et al., 2018).

The allele and genotype frequencies of the two generations (from 1947 and 2012, respectively) were calculated and statistically compared to find out any diversions that had occurred during the independent evolution. We adopted the allele frequencies of the previous generations (1947) as starting frequencies to determine the variations in the next

generations. Thus, we were able to predict the allele and genotype frequencies in the next generations, but only under ideal conditions for the population (Hardy-Weinberg Equilibrium).

MATERIALS AND METHODS

In the present pilot study, a dermatopglyphic analysis of two generations from 1947 (Markov) and 2012 (Angelova), respectively, was made. The two generations were situated in the same region of Northern Bulgaria (Fig. 1).

The sample from 2012 included 353 individuals - 300 Bulgarian Christians, 30 Bulgarian Muslims and 23 Bulgarian Turks, aged from 16 to 60. The individuals were from both genders, healthy, with no evidence of trauma on fingers. The population from 1947 included 336 individuals and was used as control group for comparison of differences in fingerprint patterns. The parameters observed were loops, arches and whorls of individual fingers of the right and left hand (Fig. 2).

Standard ink methods for taking fingerprints as described in Cummins and Midlo (1961) were used. Fingertips of both hands were impregnated with ink one by one, from I to V phalanges, and then were rolled over a white paper A4 by applying continuous pressure by the investigators.

All dermatoglyphic prints of the subjects were studied, tabulated and analyzed by applying appropriate statistical analysis. Descriptive statistics was used to summarize the data. Hardy-Weinberg equilibrium (HWE) and allele frequencies were determined. Chi-squared test, ANOVA and two-sample Z-Test was used to test the differences in fingerprints between the groups. The level of significance was set at $p<0.05$.

RESULTS

The present study included 353 subjects, 300 Bulgarian Christians, 30 Bulgarian Muslims and 30 Bulgarian Turks. The population from 1947 included 336 individuals (30 Bulgarian Muslims and 30 Bulgarian Turks) and was used as control group.

Among the groups of Bulgarian Christians from 2012 (n=300) loops showed the highest occurrence (177; 59%)

Figure 1. Geographical locations of the two generations from 1947 (Markov G. 1947) and 2012 (Angelova V.) in Bulgaria.

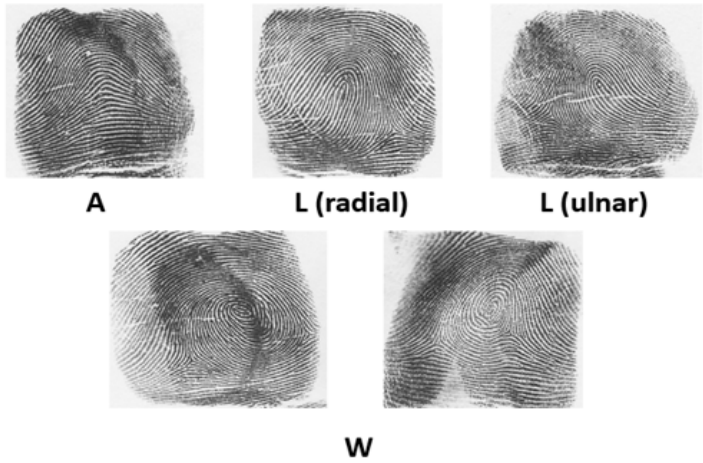

Figure 2. Different type of fingerprints recorded from the sampled Bulgarian population: A (arches); L (loops); W (whorls).

followed by whorls (101; 33.6%) and arches (22; 7.33%). Among the groups of Bulgarian Muslims from 2012 (n=30) loops showed the highest occurrence (18; 60%) followed by whorls (10; 33.3%) and arches (2; 6.7%). Among the Bulgarian Turks community (n=23) loops showed the highest occurrence (13; 56.5%), followed by whorls (7; 30.4%) and arches (3; 13.04%).

The percentage distribution of finger patterns in the two populations (from 1947 and 2012, respectively) showed that loops were the most common patterns in the three groups (56.50% - 70%). The second most common patterns were the whorls (20% - 36.4%) followed by arches (4.5% - 13.04%) (Fig. 3).

Based on the available data, we compared the proportions of each group

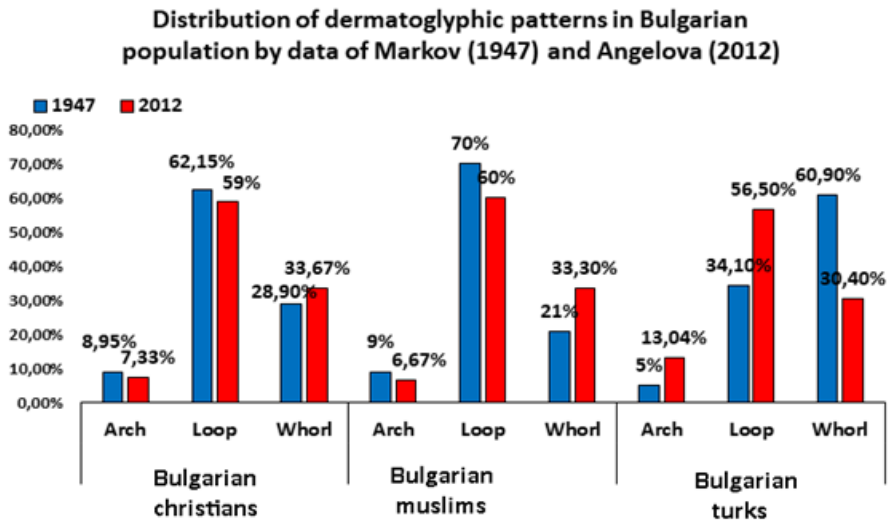

Figure 3. Percentage distribution of dermatoglyphic patterns (Arch,Loop and Whorl) in population of Bulgarian Christians, Bulgarian Muslims and Bulgarian Turks by data of Markov from 1947 (blue) and Angelova from 2012 (red).

(using count of all dermatoglyphic patterns for each group) between the two populations from 1947 and 2012, respectively. We used two-sample Z-Test for differences between proportions to determine the significant difference in dermatoglyphic patterns between groups. The results are shown in Table 1. Comparison of fingerprint proportions between each group of the two populations based on the data of Markov (1947) and Angelova (2012) showed statistical significance ($p \leq 0.05$).

The relationship between fingertip patterns of the three groups of Bulgarian population showed significant variations with respect to arches in the group of Bulgarian Turks (p=0.001) and Bulgarian Christians (p=0.03), but not in Bulgarian Muslims. Significant variations were detected in fingertip patterns between the groups of Bulgarian Christians in 1947 and 2012 with respect to loops (p=0.001) and whorls (p=0.0001). Significant variations were revealed when comparing fingerprint patterns between the groups of Bulgarian Muslims in 1947 and 2012 with respect to loops (p=0.01) and whorls (p=0.0002).

Because the population sample size was small and population variance was unknown, we used the paired sample t-test to analyze the differences in all kinds of fingerprints in groups two by two. The differences in the distribution of fingerprints patterns in the groups of Bulgarian Christians, Bulgarian Muslims and Bulgarian Turks did not reach statistical significance ($p \geq 0.05$). The results are given in Tables 2, 3 and 4.

Table 1. Relationship of dermatoglyphic patterns in population of Bulgarian Christians, Bulgarian Muslims and Bulgarian Turks between the two populations from 1947 (Markov) and 2012 Angelova).

Ethno-geographical cohorts	Pattern types	Markov, 1947		Angelova , 2012		Statistical inference
		[%]	[n*]	[%]	[n*]	
Bulgarian Christians	Arches (A)	8.80	250	7.33	220	p=0.03**
	Loops (L)	62.30	1770	59.00	1770	p=0.001**
	Whorls(W)	28.90	820	33.67	1010	p=0.0001**
Total		100.00	2840	100.00	3000	
Bulgarian Muslims	Arches (A)	10.00	30	6.70	20	p=0.144
	Loops (L)	70.00	210	60.00	180	p=0.01**
	Whorls(W)	20.00	60	33.30	100	p=0.0002**
Total		100.00	300	100.00	300	
Bulgarian Turks	Arches (A)	4.50	1	13.04	30	p=0.001**
	Loops (L)	59.10	13	56.50	130	p=0.576
	Whorls(W)	36.40	8	30.40	70	p=0.18
Total		100.00	220	100.00	230	

*n – count of dermatoglyphic patterns (fingerprints).
**The level of significance was set at $p<0.05$.

Table 2. Dermatoglyphic pattern types between two cohorts of Bulgarian Christians by data of Markov from 1947 and Angelova from 2012 (t-test).

Pattern types	Bulgarian Christians, (Markov, 1947)	Bulgarian Christians, (Angelova, 2012)	Statistical inference
Arches (A)	25	22	t= 0.08 p=0.94
Loops (L)	177	177	
Whorls (W)	82	101	
Total (N)*	284	300	

*N - total number of individuals in the sample under study.

Table 3. Dermatoglyphic pattern types between two cohorts of Bulgarian Christians by data of Markov from 1947 and Angelova from 2012.

Pattern types	Bulgarian Muslims, (Markov, 1947)	Bulgarian Muslims, (Angelova, 2012)	Statistical inference
Arches (A)	3	2	t=0 p >0.99
Loops (L)	21	18	
Whorls (W)	6	10	
Total (N)*	30	30	

*N - total number of individuals in the sample under study.

Table 4. Dermatoglyphic pattern types in cohort of Bulgarian Turks by data of Markov from 1947 and Angelova from 2012.

Pattern types	Bulgarian Turks, (Markov, 1947)	Bulgarian Turks, (Angelova, 2012)	Statistical inference
Arches (A)	1	3	t=0.07 p=0.94
Loops (L)	13	13	
Whorls (W)	8	7	
Total (N)*	220	230	

*N - total number of individuals in the sample under study.

Concerning the intergroup comparison, no significant variation was found. The distribution of dermatoglyphic traits in the three groups (Bulgarian Christians, Bulgarian Muslims and Bulgarian Turks) of the two generations from 1947 and 2012, respectively showed a similarity. Differences were not considerably great within each group. Overall, the number of dermatoglyphic patterns characterizing

the three groups was similar - the loops were the most widely observed pattern, followed by the whorls, while the arches were not numerous.

All phenotype data were used to calculate allele frequencies and probable genotypes in the two generations studied were obtained. For each group the frequencies were considered separately. We used the allelic and genotypic frequencies established in the 1947 generation as initial frequencies. In population genetics, the processes of mutations, natural selection and migrations have matter if they act over a very long period of time (thousands, millions of years). A population is in equilibrium when its state does not change without the interference of external factors. Balance can be persistent, unsustainable or neutral depending on how the population evolves from equilibrium. From the results, we can determine what the expected frequencies of A, L and W would be in the next generations. The frequencies were calculated according to Hardy Weinberg equilibrium (HWE). The allele and genotype frequencies are given in Tables 5, 6 and 7.

The phenotype data were used to calculate allele and genotype frequencies. As per the results of allele and genotype comparison, no significant variation was found. The distribution of allele and genotype frequencies in the three groups (Bulgarian Christians, Bulgarian Muslims and Bulgarian Turks) showed a similarity. The equality of allele frequencies in non-evolving populations is based on the observation that in the absence of evolution, allele frequencies in randomly breeding populations remain stable from generation to generation. From a genetic point of view, in our pilot study, the period was very short for mutations, natural selecion, migration effect and genetic drift to occur.

DISCUSSION

Dermatoglyphics characteristics have been widely used to study the variability in human populations at both the intra- and intergroup levels. They play an important role for understanding the evolution and genetic structure of human populations and the characterization of syndromes and diseases, as well as for personal identification (Fananas et al., 1996; Rosa et al., 2000; Champod et al., 2004; Noemí et al., 2016). Although there have been a few reports on the inheritance of normal fingerprint patterns, and the dominant inheritance of some fingertip features (David TJ, 1971; Herman et al., 1976), no distinct and accurate pattern of inheritance has been established. The formation of these patterns is determined by both genetic and environmental factors and they are considered polygenic traits with multifactorial inheritance (Loesch et al., 1983; Holt, 1968).

The present pilot study aimed to understand the genetic relationship among two generations - from 1947 (Markov, 1947) and 2012 (Angelova) in Northern Bulgaria using classical dermatoglyphic markers like fingertip patterns.

Our results showed that the frequencies of the main pattern types found in the analyzed sample from 2012 (Angelova) did not fall within the distribution range described by Markov (1947). We used the three general fingerprint classification types: arches (A), loops (L) and whorls (W) of the left and right hands (Aigbogun et al., 2018).The relationship of fingertip

Table 5. Comparison of allele and genotype frequencies of *A, L* and *W* in Bulgarian Christians (Markov,1947) and Bulgarian Christians (Angelova 2012).

Bulgarian Christians (Markov, 1947)			Bulgarian Christians (Angelova,2012)			Chi-square/ p-value*
Allele	N	HWE frequency	Allele	N	HWE frequency	
A	25	0.05	*A*	22	0.04	
a	259	0.95	*a*	278	0.96	p=0.54
Genotype			Genotype			
AA		0.003	*AA*		0.002	
Aa		0.095	*Aa*		0.078	
aa		0.902	*aa*		0.92	
Allele	N	HWE frequency	Allele	N	HWE frequency	
L	177	0.39	*L*	177	0.36	
l	107	0.61	*l*	123	0.64	p=0.59
Genotype			Genotype			
LL		0.15	*LL*		0.13	
Ll		0.48	*Ll*		0.46	
ll		0.37	*ll*		0.41	
Allele	N	HWE frequency	Allele	N	HWE frequency	
W	82	0.16	*W*	101	0.19	
w	202	0.84	*w*	199	0.81	p=0.57
Genotype			Genotype			
WW		0.03	*WW*		0.04	
Ww		0.27	*Ww*		0.30	
ww		0.70	*ww*		0.66	

*Chi-square test was used ($p < 0.05$).

patterns of the three groups showed significant variations with respect to arches in Bulgarian Turks (p=0.001) and Bulgarian Christians (p=0.03), but not in Bulgarian Muslims. Significant variations were detected in fingertip patterns between the groups of Bulgarian Christians with respect to loops (p=0.001) and whorls (p=0.0001). Significant variations were revealed regarding fingerprint patterns between the groups of Bulgarian Muslims with respect to loops (p=0.01) and whorls (p=0.0002). The results are in agreement with the data for the Bulgarian population as a whole as well as for some other ethnic groups.

We calculated the allele and genotype frequencies for the two studied generations

Table 6. Comparison of allele and genotype frequencies of A, L and W in Bulgarian Muslims (Markov,1947) and Bulgarian Mislims (Angelova 2012).

Bulgarian Muslims (Markov, 1947)			Bulgarian Muslims (Angelova,2012)			Chi-square/ p-value*
Allele	N	HWE frequency	Allele	N	HWE frequency	
A	3	0.05	*A*	2	0.03	
a	27	0.95	*a*	28	0.97	p=0.89
Genotype			Genotype			
AA		0.003	*AA*		0.001	
Aa		0.097	*Aa*		0.059	
aa		0.90	*aa*		0.940	
Allele	N	HWE frequency	Allele	N	HWE frequency	
L	21	0.45	*L*	18	0.37	
l	9	0.55	*l*	12	0.63	p=0.59
Genotype			Genotype			
LL		0.20	*LL*		0.14	
Ll		0.50	*Ll*		0.46	
ll		0.30	*ll*		0.40	
Allele	N	HWE frequency	Allele	N	HWE frequency	
W	6	0.11	*W*	10	0.18	
w	24	0.89	*w*	20	0.82	p=0.57
Genotype			Genotype			
WW		0.01	*WW*		0.03	
Ww		0.19	*Ww*		0.30	
ww		0.80	*ww*		0.67	

*Chi-square test was used ($p < 0.05$).

(from 1947 and 2012, respectively), considering the phenotypic data and using Hardy-Weinberg principles. We used the allelic and genotypic frequencies established in the 1947 generation as initial frequencies. Based on the polygenic inheritance analysis we determined the pattern fingerprints model and their allele frequencies in both generations. We found that with respect to the three main phenotypic traits - arches, loops and whorls, the recessive phenotypes were predominant. This fact is usual, considering the assumption that dermatoglyphic trait with no pronounced and shaped arches and whorls is predominant (Tables 5, 6, 7). The results of our pilot research showed that as part of the Bulgarian population,

Table 7. Comparison of allele and genotype frequencies of A, L and W in Bulgarian Turks (Markov,1947) and Bulgarian M (Angelova 2012).

Bulgarian Turks (Markov, 1947)			Bulgarian Turks (Angelova,2012)			Chi-square/ p-value*
Allele	N	HWE frequency	Allele	N	HWE frequency	
A	1	0.02	*A*	3	0.07	
a	21	0.98	*a*	20	0.93	p=0.60
Genotype			Genotype			
AA		0.001	*AA*		0.005	
Aa		0.039	*Aa*		0.135	
aa		0.96	*aa*		0.86	
Allele	N	HWE frequency	Allele	N	HWE frequency	
L	13	0.23	*L*	13	0.25	
l	9	0.77	*l*	10	0.75	p=0.98
Genotype			Genotype			
LL		0.05	*LL*		0.06	
Ll		0.36	*Ll*		0.38	
ll		0.59	*ll*		0.56	
Allele	N	HWE frequency	Allele	N	HWE frequency	
W	8	0.21	*W*	7	0.17	
w	14	0.79	*w*	16	0.83	P=0.91
Genotype			Genotype			
WW		0.04	*WW*		0.03	
Ww		0.33	*Ww*		0.28	
ww		0.63	*ww*		0.69	

*Chi-square test was used ($p < 0.05$).

the population in the Northern regions of Bulgaria are no exceptions in terms of dermatoglyphic patterns (Baltova and Scheil, 2007; Tornjova-Randelova S et al., 2008; Galina Yaneva et al. 1999; Karev 2008; Karev, 2011).

Comparing the allele frequencies of the two Bulgarian Christian groups from 1947 and 2012 no statistical significance was detected. In the other tested groups, such a difference was not detected as well, which may be due to the small samples or factors that affect population development such as migrations, mutations, gene flows and others. The three groups, Bulgarian Christians, Bulgarian Muslims and Bulgarian Turks occupied the same region with similar geographical, climatic and

ecological factors which makes difficult the genetic exchange as a mechanism by which new genotypes could be formed. This gives us the reason to believe that the results obtained are representative and can be regarded as reliable.

The absence of differences in allele and genotype frequencies among the three groups of the Bulgarian population can be interpreted in terms of the distribution of the population and the phenotypic expression of the different alleles not only with small representative samples.

In conclusion, regarding to the principles of independent inheritance and preservation of allele and genotype frequencies over a long period of time (in our study this period is quite short, about 70 years), our pilot study showed only small differences without statistical significance in dermatoglyphic characteristics and phenotype variations compared to those found in other regions of our country. In our opinion, the demographic and ecological factors, as well as migration and socio-economic conditions have not contributed to changes in the dermatoglyphic traits of the population.

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