

Review

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An ethnic genetics - case study of B&H: facts, fallacies and myths

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Abstract

The post-war period (1996 to present) in the Western Balkans is colored by a kind of competition among (pseudo) scientists and self-proclaimed experts in search for the deepest roots of a particular ethnic group. General conclusions have been reached based on the distribution of a single or only few genetic markers, with no reference to the specific pheno-genotype system studied. The conclusions were all biased by earlier misconceptions and myths about the successive colonization of the Balkans and the inter-genetic relationships among regional populations. In this paper we elaborate methodology and limitations and misconceptions that arise from unsubstantiated use thereof.

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Introduction - Population genetics in B&H timeline

Since its onset, the anthropogenetic research in the Balkans has been carried out in several stages with ever increasing levels of complexity and comprehensiveness. The first demographic-genetic surveys in B&H were published in the last decade

of the 19th century by two Austrian military physicians with the rank of "hauptmann" Heinrich Himmel (Herzegovina recruits) and "obersturmarzt" Augustin Weissbach (B&H recruits) (Weissbach, 1895). The following stage was featured by the analysis of individual phenotypic systems with

basic analysis of gene and phenotype frequencies. Periods 1934-1935 and 1953-1966 were dedicated to the analysis of ABO, Rh and MN systems (Kalić and Kostić, 1934; Bošković 1965, Berberović 1968). In 1967-1976 populational studies advanced with respect to the number of markers observed and the level of complexity of mathematical models applied. Numerous biochemical-physiological and morphological characteristics were observed in predominantly urban (sub)populations. Complex study of interpopulational genetic distance was introduced in 1977 with a shift of focus from urban to predominantly non-migratory rural populations (Hadžiselimović, 1981, 1983, 1984).

At the end of this research cycle, the possible relationships between the observed indicators of genetic distance and its possible causes were also examined. This is primarily related to the degree of isolation of local populations. The frequency of marital distance „zero“ (marriage couple originated from the same local population), distribution of the mean marital distance, endogamy coefficient, differential reproductive rate, population size, and correlation between mean geographical distance and mean genetic distance. The highest positive (significant) correlation ($p < 0.05$) was observed between genetic distance and population size. However, the largest (significant also) negative correlation was observed between the average genetic distance and marital distance „zero“ (0). The overall results indicate that the studied populations constitute „inland islands“ in the sense that the married couples originated from the same local community in 90-98% (Hadžiselimović and Zovko, 1987). From 2002 onwards, population studies include direct genetic markers (nuclear and mitochondrial). The largest dataset ($N =$ about 20,000) was collected for the purpose of DNA profiling of the remains of war victims and consisted of STR profiles of exhumed victims and reference profiles of their surviving relatives. Genetic distance among B&H local and metapopulations, with regard to several (nuclear and mitochondrial) genetic markers was extensively studied by numerous authors (Marjanović et al. 2005a, 2005b, Battaglia et al 2009, Pojskic et al 2013a, 2013b, Ahmić et al. 2014, Kapur-Pojskic et al. 2014, Kovacevic et al., 2014, Sarac et al., 2014, Carreras-Torres et al., 2016,

Babić et al., 2017, Pilav et al, 2017, Čakar et al. 2017, Ahmic et al, 2018). Since 2016, contemporary research has gradually been expanding into the field of omic technologies. Following positive experiences in genome analyses (genomics) the first results of transcriptome analysis are expected soon.



Figure 1. Eye color distribution in B&H populations (blue, dark and grey-brown), the last decade of the 19th century (Weisbach, 1895)

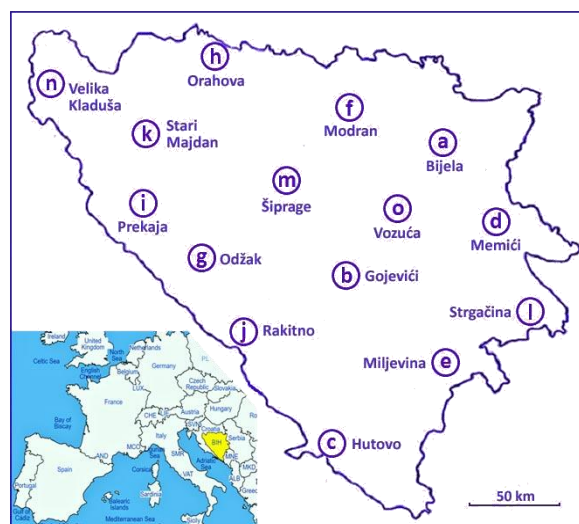


Figure 2. Spatial distribution of 15 local B&H populations studied in the period 1973-1976

Facts

Previous population-genetic studies of classical morphological, biochemical-physiological traits and molecular-biological markers have not shown any differences in the genetic structure among ethnic populations in Bosnia and Herzegovina. It was

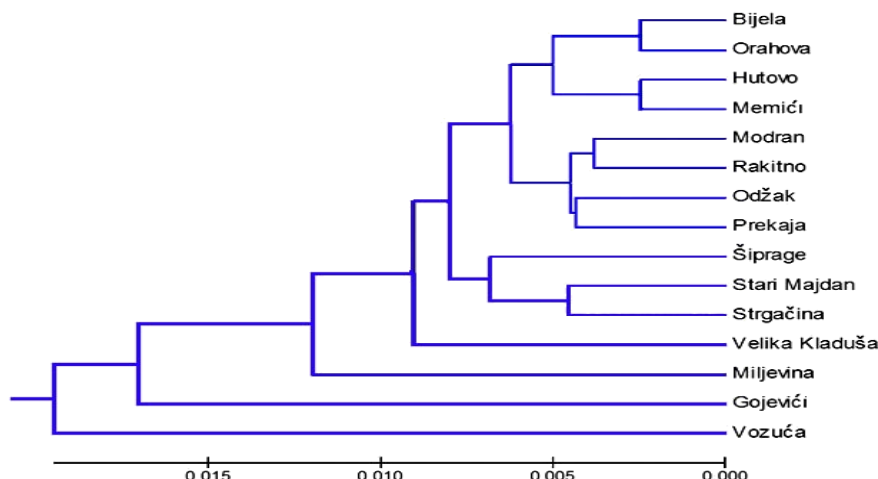


Figure 3. Genetic distance among 15 local B&H populations based on 15 phenotypic markers, i.e. the corresponding alleles (ABO blood groups (lo) ABO(H) antigens secretion (se), red (cvp) and green (cvd) color vision, PTC testing (t), tongue rolling (r) and fissuring (f), ear lobe types (l), chin dimple (ch), midphalangeal hairiness (d), crooked 5th finger (rcf), high-hicker's thumb (dht), proximal thumb joint hyperextensibility (pht), digital index (lf), and long hand nails(ln))

also noted that the differences among local populations within particular ethnic group exceed or are equal to their distance from the said ethnic group as a whole. The latest data, collected through the popular and massive worldwide MyHeritage project (<https://www.myheritage.com/>), has shed more light on the existing ideas about possible similarities and differences between Balkan populations and ethnic groups. Pojskić et al. (2013) studied the nuclear Alu markers, and reached similar conclusions. Interestingly though, when gender-specific markers (Y-chromosome) were analyzed, the conclusions were still the same (Marjanović et al. 2005a, 2005b). In addition, Pojskić et al. (2013) and other studies (Ahmić, 2005) have established that the net result of genetic diversity study of populations included largely depended on the complex of studied markers. On the other hand, the same authors also concluded that genetic distance estimation heavily depended on the mathematical model used in the evaluation of the observed measures.

Fallacies - Neglected starting points

“Genetics of Nations“/ Ethnic genetics is a popular syntagma which has entered the scientific vocabulary as well. It is completely unnecessary and has enormous potential for misuse in Western Balkans region. Recent interpretations were based

on erroneous premises such as:

- Axiomatic – uncritical acceptance of consensus Ex-Yugoslavian historiography;
- Complete extinction of ancient population and all earlier epochs;
- Complete replacement in the succession of landmark immigration in B&H;
- Population discontinuity of present-day B&H territory;
- Identification of linguistic and other cultural changes with the biological essence of B&H population.

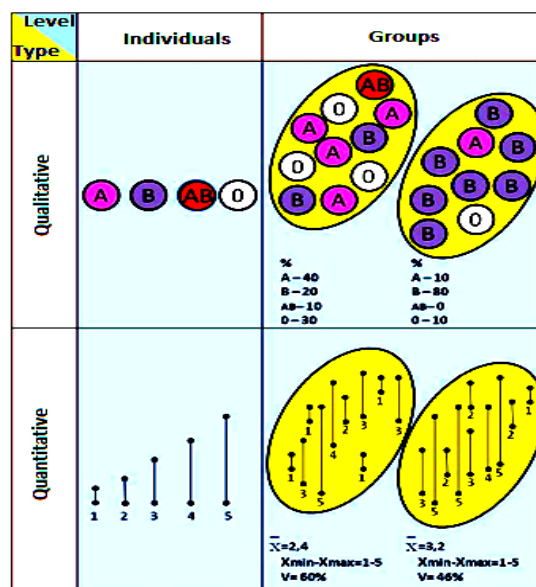


Figure 4. Basic parameters of the individual and population differences regarding the qualitative and quantitative variation (Hadžiselimović, 2019)

Striking conclusions are extracted from genetic distance based on a single or few genetic markers.

Human individual diversity rests on variation of extremely large number of qualitative and quantitative traits with virtually limitless possibilities to combine their variants (phenotypes). Even in small group of human individuals, it would be difficult to find two individuals with the same variant of only one trait; even fewer that are matched according to two traits, while those that possess the same combination of any three traits are extremely rare. The number of combinations of variants of dimorphic traits increases according to the geometric progression with 2 as a base (= number of alternative variants) and n as an exponent (= number of traits included). Thus, when combining two traits the number of combinations is 4 ($= 2^2$), three traits give 8 combinations ($= 2^3$) while four traits result in 16 ($= 2^4$) different individual combinations. At the point when we combine 34 traits the number of combination reaches 2^{34} or over 17 billion, more than 2.5 times the total number of modern inhabitants of the planet, i.e. 1/5 of all people who have seen the light of day on Earth so far. One hundred such properties give the number of combinations far greater than not only the total number of people born so far, but all living beings together. If we factor in the plasticity of genotypes in different environmental scenarios and ontogenetic variability, the number of possible phenotypic combinations in the human population exceeds the total count of electrons in the Cosmos.

Group or population diversity, although derived from the individual biological differences, represents a new quality with higher level of organization and complexity. It has specific manifestations and measures; regardless of whether we consider qualitative or quantitative individual variation, group - specific properties inevitably have quantitative (quantitative) parameters. Individuals differ according to the variant or category of particular qualitative or quantitative trait while intergroup differences are commonly defined by the quantity of individual phenotypes of the observed trait. If we take an example of ABO system of blood groups two individuals may differ in the alternative quality (biochemical structure) of ABO antigens so that one individual is group A while the other is group B.

However, two hypothetical populations differ in relative frequency of particular blood group so one group contains 40% individuals with blood group A while the other contains 60% of the same variant.

This coarse indicator does not provide even the most elementary information on the intra-group variation of the individuals involved, so the same amounts can occur in extreme cases (absolute homogeneity or heterogeneity). Occurrence of two biologically identical individuals in all of humanity throughout history is infinitely unlikely, thus occurrence of two biologically identical human groups is theoretically impossible.

The genetic characteristics of a human population may rest on:

- the presence of one or more allelogens that are not present in other populations;
- the complete absence of alleles that occur regularly in other populations;
- both of these phenomena;
- general presence but significantly different frequency of individual allelogens in all populations compared.

Neither of the group properties applies to its individual members.

Myths

Certain ideas about the sequence of settling the area have been propagated for centuries and are consequently deeply rooted in the Balkans.

With this respect, official theories on mass migrations and resettlement have been developed, which have also influenced the official social and political atmosphere. The most prominent tales that reached the level of a myth are:

- Complete Bogomilisation of the parent population (Constituents of AllBosnian Church were erroneously recognized as Bogomils because of dualistic concept of belief);
- Predominant Slavisation of B&H population;
- Existence of a defining genetic marker(s) of a "national/ethnic group" that clearly differentiates a given group from related ethnicities, neighbors and the rest of humanity.

The Balkans is a fertile breeding ground for myths. However, the best historically ingrained myth is the

Serbian one about the „heavenly people“. Immediately following the Kosovo battle popular epic poetry emerged that glorified the heroes and vilified the traitors. In the former Yugoslavia (the first of 1918 and the second, socialist one of 1945), this myth was also imposed in all federal units. That syntagma is comparable to the famous Nazi racist thesis on superhumans (*Übermensch*).

Some authors argue that this myth has been developing spontaneously with no interference, until the genocide in Bosnia and Herzegovina. There is also an indication that the Serbian leadership used the myth for political purposes to channel military and paramilitary formations in the direction of ethnic cleansing and war crimes (Anzulović, 1999).

During the War against Bosnia (the most frequently used euphemism is „Bosnian War“), the myth was encouraged by certain Bosnian Serb leaders to describe Bosniaks as "genetic junk" (Statement by Biljana Plavšić given to Belgrade's academic community in 1992. Only Nikola Tucić, academician responded, qualifying the statement as a classic form of racism).

Facts vs Fallacies and Myths

Analyzing biochemical-physiological characters (BFC), static-morphological characters (SMC) and dynamic-morphological characters (DMC), different clusters were created (Figures 5-10).

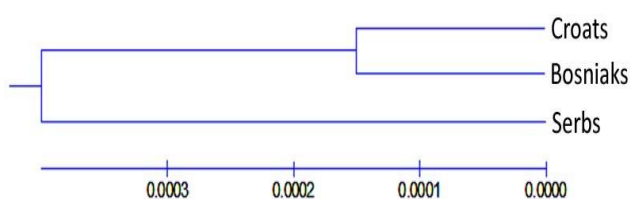


Figure 5. UPGMA dendrogram based on the genetic distance analysis according to Reynolds et al. (1983) Subpopulations in B&H: BFC

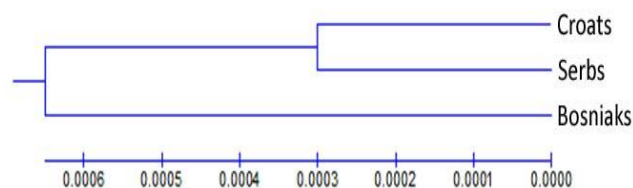


Figure 6. UPGMA dendrogram based on the genetic distance analysis according to Reynolds et al. (1983) Subpopulations in B&H: SMC

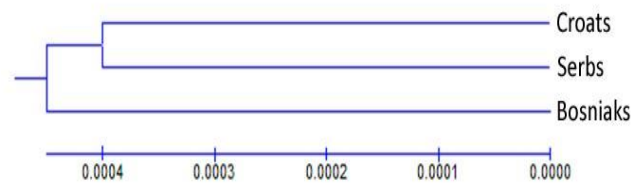


Figure 7. UPGMA dendrogram based on the genetic distance analysis according to Reynolds et al. (1983) Subpopulations in B&H: Overall characters observed

The three dendrograms observed exhibit markedly different clustering according to the complexes of the studied traits (figures 5-7). The largest difference with other complexes is shown in the observed group of the static-morphological properties.

Differences in clustering according to different trait complexes were also observed by Ahmić (2005). She found that the complex of dynamic-morphological traits deviates the most from the other two trait complexes: biochemical-physiological and static-morphological (figures 8-10).

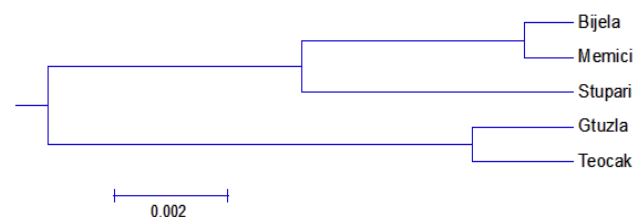


Figure 8: Genetic distance relationships with respect to the complex of BFC among the compared populations in Tuzla Region (Ahmić, 2005)

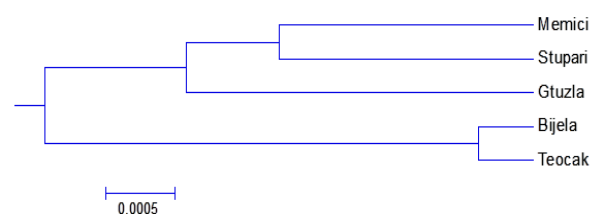


Figure 9. Genetic distance relationships with respect to the complex of SMC among the compared populations in Tuzla Region (Ahmić, 2005)

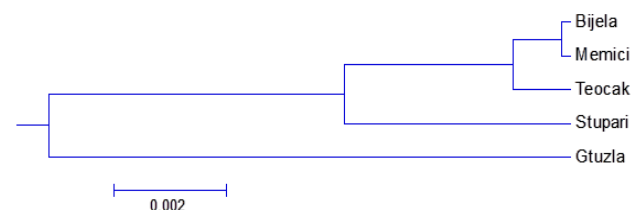


Figure 10. Genetic distance relationships with respect to the complex of DMC among the compared populations in Tuzla Region (Ahmić, 2005)

The latest findings on the MyHeritage Platform (with millions of family DNA profiles) have revealed new genetic evidence that does not call into question the indigenous origins of Albanians, Bosniaks, Montenegrins, Serbs, Croats and Macedonians. It has been proven time and again that popular legends about the oldest nations in the Balkans are pure misconceptions, lies and / or superstitions. According to direct genetic markers, the closest ties share Bosniaks, Croats and Serbs, and close to them are Hungarians, Romanians, Bulgarians, Czechs, Slovaks, Poles, Austrians, Greeks, Italians, Germans while Russians, Moldovans and some others populations are only distantly related.

The decades-old myth of the great migration of the Slavs into the Balkan wasteland is overthrown because the so-called "Slavic gene" is slightly expressed in all present-day nations, with relative frequency in:

- Serbs: 30%,
- Croats: 20% and
- Bosniaks: 15%.

According to research conducted by Al Jazeera (2017) Bosnians and Herzegovinians are believed to inherit the remnants of the gene pool of:

- Illyrians: 40%,
- Germans: 20%,
- Celts: 15%, a
- Slavs: 10% .

The population of B&H was not originally constituted in the Slavic but in the Illyrian era, with genetically related haplogroups very similar to Croats and Bosniaks. According to these haplogroups, Turks are twice as close to Albanians as to Serbs and Macedonians, with Bosniaks being the most further of the four populations. No nation in the Balkans has a unique, distinctive haplogroup. That is true for any other European population.

Figure 11, however, clearly shows indicative subclustering (Bosnia and Herzegovina, Slovenia, Austria, Hungary, and Croatia), which particularly singles out a group of countries of fairly heterogeneous geographical, as well as ethnic origin (Pilav et al., 2017).

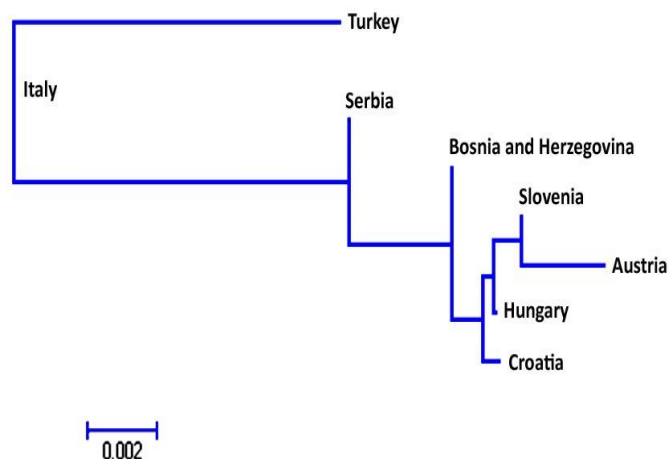


Figure 11. UPGMA genetic distance among selected European population, based on STR markers

Conclusions

The present-day population of BiH inherits an indigenous gene pool with more or less pronounced "genetic memories" of external invasions, from prehistory and ancient times to the recent epoch. This is not to contradict the fact that it has – in whole or in part – been affixed to various socio-political or appropriate professionally adapted labels. The estimated genetic distance depends primarily on the number and nature of the genetic markers selected, as well as patterns used. Even when the analysis of relationship among the Balkans peoples involved impressively large samples and genetic markers complexes, the general conclusions were drawn overconfidently. Similar attempts were made based on single or few markers with no comment that the presented conclusions exclusively pertain to the studied genetic indicators in the studied samples. Identification of particular gene or genetic marker that would be highly specific for particular nation is both theoretically and practically impossible. Furthermore, there are few markers that cannot be found in most populations.

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