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Original scientific paper

ANTHROPOGENETIC VARIABILITY IN GROUPS OF CHILDREN FROM REGULAR AND SPECIAL SCHOOLS FROM DIFFERENT LOCALITIES IN SERBIA

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This population-genetic study compares morpholophysiological and genetic variability in five control groups of individuals (children from five regular schools, N= 996) with children from that many special schools (N= 736) from Serbia, by using a test of determination of homozygously recessive characteristics in humans (HRC-test). Genetic homozygosity degree showed not only statistically significant difference between the mean values obtained for two groups of studied samples (control group 6.95± 0.07; children from special schools 8.63± 0.08 HRCs, out of 30 analyzed characteristics), but also differences in the type of distribution, as well as the presence of specific combinations of such traits. Results of comparisons done in different places (Kraljevo, Nis, Vranje, Leskovac, Pirot) showed the same tendency- the increase of genetic homozygosity and relative decrease of variability in samples of children from special schools. The number of HRCs among individuals from control groups varied from 2 to 15/30, and from 3 to 16/30 among children from special schools. It is possible that increased recessive homozygosity present in the group of children from special schools leads to increase of genetic loads, what may cause easier expression of some physiological and mental abilities that children from special schools have.A great individual variation in amount of genetic homozygosity that exists among human individuals may influence their potentials for different kinds of adaptation, including their mental abilities, physical capacities or resistance to different diseases.

Key words: genetic homozygosity, anthropogenetic variability, school children

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INTRODUCTION

Since only a small number of loci with allelic genes determining an exact biochemical process are known, estimating genetic homozygosity in humans is complex and sensitive task. Using *Drosophila-model*, MARINKOVIC (1967) took a part in 1960s and 1970s, in world-wide studies of "genetic loads", which contributed to our understanding of the maintenance of genetic variation in natural populations. Provided the type of inheritance and variability is carefully studied, it can be seen also in humans that a series of morpho-physiological traits are controlled by one or several genes. Making use of this information, several authors of Belgrade population-genetic school have studied the distribution and frequency of a series of such homozygously expressed recessive morphophysiological characters ('HRCs') in order to estimate individual and group difference in their presence (e.g. among ill versus healthy individuals, pupils from special and regular schools, carriers of different blood types, etc.). With gradual improvements, these studies last more than two decades (BLAGOJEVIC *et al*., 1989; MARINKOVIC *et al.*, 1990, 1994, 2008; MARINKOVIC and CVJETICANIN, 1991, 2007, 2013; MARKOVIC-DENIC *et al*., 1992; MARJANOVIC *et al*., 1992; RISTIC *et al.*, 1992; TOMIC *et al*., 1994; CVJETICANIN and MARINKOVIC, 2005a, b, 2009; PESUT, 2004: BRANKOVIC, 2006; PESUT and MARINKOVIC, 2009; CVJETIĆANIN, 2000, 2015).

It is established that certain morpho-physiological characteristics are expressed as qualitative traits and that they are under the control of one or small number of genes located on different human chromosomes. Thus, they could be considered as genetic markers of appropriate chromosomes, as well as of numerous surrounding genes controlling different fitness properties. Genes which determine expression of certain phenotype traits along with environmental factors could potentially influence, to the certain degree, the development of certain diseases, or different predispositions. The amount of recessive homozygosity established by HRC-test is practically an estimation of genetic loads present in any specific sample of humans.

Complex traits in humans have polygenic basis of determination of individual variation, when a proportion of loci which are in homo- or heterozygous states is crucial for expression of particular characters. Taking all this into account, our assumption was that groups of children from regular and special schools may differ in genetic homozygosity and variability, what will contribute to our untertanding of population-genetic differences eventually expressed in these studies.

MATERIALS AND METHODS

For the estimation of the degree of recessive homozygosity we used "HRC-test", established in late 1980s by professor D.Marinkovic at Department of genetics and evolution, Faculty of Science in Belgrade, and applied in humans. This test includes in our study thirty selected and approved homozygously recessive characters (HRCs), where extreme appearance of tested traits was expressed as homozygous state of either individual or of a group of genes. The HRC-test has been developed to establish the proportion of clearly expressed homozygously recessive morphophysiological characters in every individual, as markers of their chromosomal homozygosity, implicating the degree of genetic loads in humans with their cumulative effects. It is a highly suitable method for estimating individual homozygosity by direct observation, as it takes only a few minutes to analyse the presence of 20-30 defined phenotype characteristics (Table

1). OMIM numbers and eventual gene location of some of observed characteristics are given also in this journal (CVJETICANIN, 2015).

In this study comparative analyses were made by the same person, with equal criteria for determining extremely pronounced homozygously recessive characters in tested groups of observed individuals.

Variations in presence of analysed characters were estimated applying standard statistical procedures, and by comparing the means, variances, and the distribution shapes between two groups of observed school children. Commom tests to determine the differences in the mean values, scope and type of variability (t-test, S^2, and X^2, respectively) have been used. Presence of the studied genetically controlled recessive characters was used as a parameter for homozygosity of corresponding genes and chromosomes. Based on the criteria related to achievements, the subjects were devided into the following categories:

SS (N = 736) – children from special schools (8 to 15 years of age; IQ $\leq$ 80)
SS Kraljevo **(**N= 88 pupils)
SS Nis **(**N= 215 pupils)
SS Vranje **(**N= 95 pupils)
SS Leskovac **(**N= 158 pupils)
SS Pirot **(**N= 180 pupils)
RS (N=996) – children from regular schools (randomly chosen individuals belonging to the same age groups and locality)
RS Kraljevo **(**N= 176 pupils)
RS Nis **(**N= 215 pupils)
RS Vranje **(**N= 190 pupils)
RS Leskovac **(**N= 158 pupils)
RS Pirot **(**N= 257 pupils)
All Groups of tested individuals belong to the same ethnic group (Serbian population).

RESULTS

When examining the HRC frequency distribution in the groups of children from special and regular schools (Table 1), it is obvious that the mean value of HRCs in the total sample of individuals from special schools is significantly higher than in the control group of pupils from regular schools (RS: 6.95± 0.07; SS: 8.63± 0.08; $t = 4{,}33$, $p < 0.001$).

As for the distribution of HRC frequency among the members of special and regular schools, it can be seen that HRCs in the group of individuals from special schools have a tendency towards higher values, suggesting that different genetic dispositions at the polygenic level may exist between the two tested samples (Figure 1). The number of HRCs among individuals from control groups varied from 2 to 15 and among children from special schools from 3 to 16.

From the data presented in Table 1 it can be seen that almost all tested recessive characteristics are expressed to a greater degree in the group of special school children, and in five of observed characters increase in recessive homozygosity is statistically significant. The type of individual variation in two studied groups was also significantly differed ($\Sigma\chi^2$ = 89.42***), showing that their genetic dispositions could be remarkably different.

Table 1. The frequencies of HRC among groups of special schools and individuals of control sample in Serbia

Homozigously recessive traits	RS % N = 996	SS % N = 736	χ^2
1. Flat scalp	52.8	57.5	0.41
2. Strait hair	51.8	56.0	0.33
3. Blond hair	14.8	19.3	1.32
4. Soft hair	19.9	23.4	0.57
5. Double top of the hair	1.7	3.5	5.65*
6. Reverse top of the hair	1.2	2.9	2.25
7. Thin eye-brow	12.7	16.6	1.21
8. Blue eyes	21.1	25.1	0.77
9. Freckles	14.2	17.0	0.56
10. Dimple in cheek	21.6	23.2	0.12
11. Abnormal ear shape	11.2	15.8	1.81
12. Ear without Darvin's lump	51.7	60.5	1.48
13. Attached ear lobe	28.2	38.0	3.42
14. Face asymmetry	1.2	5.7	16.83***
15. Small nose	12.2	13.7	0.17
16. Narrow nostrils	22.2	26.2	1.42
17. Small lips	8.1	11.5	1.34
18. Thin lips	25.4	31.3	6.52*
19. Small teeth	12.4	21.5	0.55
20. Yellow teeth	27.6	31.5	6.42*
21. Inability to tongue curving	37.2	52.7	3.27
22. Inability to tongue rolling	34.2	44.8	9.44**
23. Left handedness	9.4	15.2	0.45
24. Index finger longer than 4th finger	38.4	42.5	3.12
25. Right thumb over left thumb	45.8	57.7	2.96
26. Thumb hiperextensibility	15.2	21.9	2.67
27. Three strings in the wrist	19.8	28.4	3.75
28. Absence of mid digital hair	45.3	63.0	6.85**
29. Nail abn.	14.5	20.9	2.89
30. Small finger abn.	13.9	20.1	2.81
		$\Sigma\chi^2$ = 89.42***	

*p<0.05 **p<0.01 ***p<0.001, Control (RS) N=996 $\overline{X}_{hrc/30}$ = 6.95± 0.07

Special schools (SS) N=736 $\overline{X}_{hrc/30}$ = 8.63± 0.08 t = 4,33, p < 0.001

Genetic load' RS/SS = 1 - 0,74±0,03 = 26 + 1%

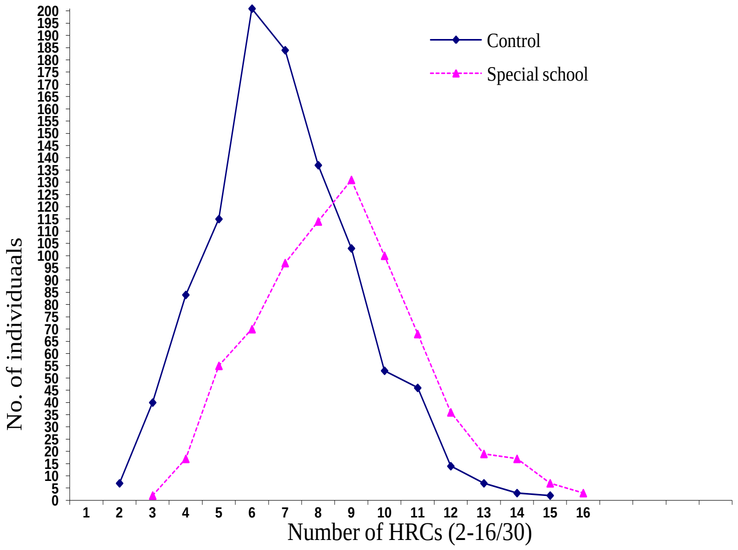

Figure 1. Distribution of 30 qualitative morphophisilogical traits in the groups of children from special and regular schools

Control N=996 $\overline{\mathbf{X}}_{hrc/30} = 6.95 \pm 0.07$

Special schools N=736 $\overline{\mathbf{X}}_{hrc/30} = 8.63 \pm 0.08$ $t = 4{,}33$, $p < 0.001$

There are no differences between localities among groups of children from special schools, nor among those from regular schools. But each group of pupils from special schools from Vranje, Leskovac, Nis, Pirot and Kraljevo showed a significant difference in mean values of genetic homozygosity, and in genetic variability, compared in the same place to appropriate control group from a regular school (Table 2). All tested groups of pupils from special schools showed an increased genetic homozygosity as well as genetic variability, for tested recessive traits.

Table 2.The presence of homozygous recessive characteristics based on the study of 30 qualitative morpho-physiological traits in the groups of regular and special schools children from different places.

	Regular schools	Special schools	t-test	*F*-test
Complete sample	N=996 $\overline{X}_{hrc/30} = 6.95 \pm 0.07$ 23%±0.5 V=31,79%	N=736 $\overline{X}_{hrc/30} = 8.63 \pm 0.08$ 29%±0.6 V=27.80 %	4.33^{**}	1.17^{**}
Vranje	N=190 $\overline{X}_{hrc/30} = 6.91 \pm 0.03$ 23% V=35.74%	N=95 $\overline{X}_{hrc/30} = 9.05 \pm 0.26$ 30.3% V=28.6 %	3.97^{***}	1.09^{**}
Leskovac	N=158 $\overline{X}_{hrc/30} = 7.15 \pm 0.15$ 24% V=27.97 %	N=158 $\overline{X}_{hrc/30} = 8.95 \pm 0.1$ 29,7% V=25.36%	3.13^{**}	1.28^{**}
Niš	N=215 $\overline{X}_{hrc/30} = 6.53 \pm 0.13$ 21.2% V=30.78 %	N=215 $\overline{X}_{hrc/30} = 8.05 \pm 0.16$ 27% V=29.93 %	2.86^{**}	1.43^{**}
Pirot	N=257 $\overline{X}_{hrc/30} = 7.19 \pm 0.15$ 24% V=32.96 %	N=180 $\overline{X}_{hrc/30} = 8.85 \pm 0.18$ 29,7% V=26.66 %	3.09^{**}	1.00^{**}
Kraljevo	N=176 $\overline{X}_{hrc/30} = 6.88 \pm 0.14$ 23% V=27.18 %	N=88 $\overline{X}_{hrc/30} = 8.69 \pm 0.22$ 29% V=24.51 %	3.01^{**}	1.09^{**}
Average homozygosity	21.2-24%	27-30.3%		

$^{*}p<0.05$ $^{**}p<0.01$ $^{***}p<0.001$

DISCUSSION

According to the data presented in this study, the frequency distribution of the tested HRCs was different in compared groups of individuals. Average homozygosity in RS amounted 23±0.5%, whereas in SS it was 29±0.6%, manifesting a population genetic difference present between them (Tables 1 and 2). In these comparisons, characteristic groups of traits were present in a different degree among children from regular and special schools, suggesting a correlation between different combinations of polygenes which may be involved in expression of specific physical and psychological abilities (Table 1). Those findings are in correlation with the results of some of previous studies of genetic homozygosity in the groups of children from special and regular schools from Belgrade (MARINKOVIC *et al*., 1990).

The average proportion of HRCs in our present control group of individuals is 23%, what is in correlation with results for control samples in numerous studies done before in Serbian populations (CVJETICANIN and MARINKOVIC, 2005b; BRANKOVIC, 2006; MARINKOVIC, *et al*., 2008, CVJETICANIN, 2015). No information on potential differences among ethnic groups in the estimated degrees of homozygosity is available, but relatively small variation among local populations does exist (BRANKOVIC, 2006; MARINKOVIC and CVJETICANIN, 2013), what our results from different places of Serbia also show (average homozygosity varies from 21.2% to 24%, Table 2).

As the HRC test covers homozygosity of all chromosomes in general, it may happen that specific genes, discovered to be responsible for expression of some physical and psychological properties, could be located at some of these chromosomes and that expressivity of those genes may depend on the status of homozygosity of other loci.

The increased degree of genetic homozygosity and relatively decreased variability for tested genes in the samples of children from special schools, may be also a result of the pleiotropic effects of genes responsible for the expression of some morpho-physiological characteristics and mental abilities. Those genes would determine the expression not only of specific morpho-physiological and mental abilities, but also a group of other characteristics, including some HRC properties as well (MARINKOVIC *et al.*,1994; CVJETICANIN and MARINKOVIC, 2005 a,b, 2008, 2009, 2013).

The fact that in these comparisons genetic homozygosity was significantly increased in the samples of special school children (compared to the groups of children from regular schools), as markers of other numerous loci, gives an impression that genetic loads, which may cause diminished body resistance, are substantially present in such samples of human populations. Calculated in Table 1 from SS/RS ratios, genetic loads amounted 26 + 1%.

Relatively large individual variation in the studied HRCs (from 2 to 15 in the control group and from 3 to 16 among children from special schools; Figure 1), covering almost all parts of the human body, is also informative of how large a variation in genetic homeostasis may exist in human individuals, with a higher chance of extreme genotypes being exposed to the risk of suffering from specific metabolic and developmental malformations.

Consequently, the future application of HRC testing can be valuable for predicting some extremely deviant genotypes, possibly more susceptible to different dispositions or diseases. Such individuals could be properly treated much before the appearance of possible malformations, since they are also a part of a biologically wide human variation.

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ANTROPOGENETIČKA VARIJABILNOST U GRUPAMA DECE IZ REGULARNIH I SPECIJALNIH ŠKOLA SA RAZLIČITIH LOKALITETA U SRBIJI

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Izvod

Koristeći test za utvrđivanje homozigotno recesivnih osobina kod ljudi (HRO-test) u ovoj populaciono-genetičkoj studiji upoređivana je morfofiziološka i genetička varijabilnosti kod dece iz regularnih škola (RS, N=996) sa decom iz više specijalnih škola (SS, N=736).Rezultati naše studije pokazuju prisustvo zanačajnih razlika kako u prosečnim vrednostima genetičke homozigotnosti (RS- 6.95± 0.07; SS- 8.63± 0.08 HRO, od 30 testiranih karakteristika), tako i u distribuciji i varijabilnosti u prisustvu specifičnih kombinacija testiranih osobina. Ovi rezultati upućuju na kompleksnu poligensku razliku među testiranim grupama ispitanika. Rezultati poređenja sa različitih lokaliteta (Kraljevo, Niš, Vranje, Leskovac, Pirot) pokazuju istu tendenciju - uvećanje genetičke homozigotnosti i relativno smanjenje varijabilnosti u uzorcima dece iz specijalnih škola. Moguće je da ovo uvećanje recesivne homozigotnosti prisutno u grupi dece iz specijalnih škola vodi ka uvećanju genetičkih opterećenja koja bi mogla dovesti do ispoljavanja nekih fizioloških i mentalnih sposobnosti koju deca iz specijalnih škola imaju. Velike individualne varijacije u stepenu genetičke homozigotnosti koje postoje među ljudima mogu uticati na njihov potencijal za prilagođavanje, uključujući njihove mentalne sposobnosti, fizičke kapacitete ili otpornost na različite bolesti.

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