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Medical Sciences

Characteristics of the Frequency Distribution of some Immunogenic Markers in the Population of the Mountain Region (Khulo) of Ajara

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ABSTRACT. Red blood group antigens represent a genetically stably determined trait, being of many-sided biological and clinical significance. The indigenous Ajarian population (105 subjects) was investigated for ABO, Rh-Hr, Kell, MN red blood group markers. Using immunoserologic methods, 13 erythrocytic group antigens (two of the ABO system: A, B; seven of the RH system: C, C, C^w, D, D^u, E, e; two of the Kell system: K and k, and two of the MN system: M and N antigens) were studied. The obtained results were statistically processed. 44±4.9% of the said region's population are carriers of the blood group 0(I), 37±4.82% are carriers of the phenotypic group A(II). Rather high is the ratio of the B(III) blood-group carriers (17±3.7%). The AB(IV) blood group is observable in 2±1.4%. In studying individual antigens of the Rh system in the Khulo population, the maximum frequency distribution of the *e* antigen was found (100±3.12%), being followed in frequency by the *c* antigen (89±3.12%). The frequency of the *D* antigen made up 82±3.8%, and that of the *d* antigen - 18±3.8%. The frequency of the *c* antigen made up 61±4.8%, that of the *E* antigen 23±4.2%. Six Rh-phenotypic groups of various frequencies were recorded for the Khulo region population. The CcDee phenotype is the most characteristic of the said region's population. Its distribution frequency amounts to 38±4.8%. The major part of the population is the carrier of the M-phenotypic group. The ratio of the MN phenotypic group carrier is 1.5 times less (32±4.6%). The N phenotypic group is characterized by a relatively lower frequency (20±4%). *© 2009 Bull. Georg. Natl. Acad. Sci.*

Key words: *antigen, phenotypic groups, allele.*

Introduction

The red blood group antigens represent genetically stably determined features [1, 2]. In spite of the stable specific traits, the respective hereditary factors are characterized by rather high polymorphism at the level of populations and species within the species' gene pool. This in itself is indicative of the essential significance of the phenotypic individuality determined by different gene combinations in the establishment of a common adaptive balance with the respective genotype environment. Hence, the erythrocytic antigenic trait is of many-sided biological and clinical significance [3-6].

The principal bio-clinical significance of the erythrocytic group antigens is still associated with the living immune characteristics. It plays a special role in blood transfusion [3], epidemiology [4] and transplantology [5,6]. Distinguished is the significance of the said systems in human genetics [8, 9], and particularly in terms of studying its population peculiarities [10-13].

The erythrocytic group systems have special significance in ethnical anthropology [11, 14-17]. Their

hereditary bases are so stable that their study in order to identify the origin of a specific ethnical group will provide reliable data [15,16,18]. Humans are so individual by their group antigens that they can serve as their identity identifiers. Based on the said property, blood group systems are widely used in forensic medicine [19, 20] and criminology.

As it seems, the composition of the blood group systems' antigens in human populations is a result of a balance polymorphism established during the evolutionary time periods. According to literary sources [21-25], a correlation between the balance polymorphism and various infectious or non-infectious diseases has been established by the erythrocytic group antigens.

Proceeding from the above, we have set ourselves an objective to establish the genetic geography of the erythrocytic group antigens in the highland Khulo region of Ajara. The available data concerning the distribution of the erythrocytic group antigens in the Ajarian Autonomous Republic are rather scarce.

Materials and Methods

The indigenous Ajarian population of the Khulo region (105 subjects in total) have been studied for the ABO, Rh-Hr, Kell, and MN erythrocytic group markers. Using immunoserologic methods, we investigated 13 erythrocytic group antigens in blood (two of the ABO system: A, B; seven of the RH system: C, C, C^w, D, D^u, E, e; two of the Kell system: K and k, and two of the MN system: M and N antigens).

In the course of the work the following specific test systems were used: anti-AB, -B, -A, -D, -CD(G), -C, -c, -E, -Ce, -e, -K, -M, -N (Gemostandart Ltd., Moscow), standard 0(I), A(II), B(III) group erythrocytes and standard 0(I), A(II), B(III), AB (IV) serums (the titer of the used reagents was not lower than 1:32).

The obtained results were statistically processed. The ABO system gene alleles' frequency was computed by the formula proposed by F. Bernstein and used in investigation of three-allele genetic systems. The frequency of the 0, A and B genes in the given case will be indicated by the letters ***r***, ***p*** and ***q***:

$r = \sqrt{O}$;

$p = 1 - \sqrt{A+O}$,

$q = 1 - \sqrt{B+O}$,

where 0, A and B – 0(I), A(II) and B(III) is the ratio of the group carrier people in relation to the total number of the subjects of the study.

The frequency of the *Rh*-system genes and haploid types was computed by using the following formulas:

1. $D = 1 - \sqrt{dd}$;
2. $C = 1 - \sqrt{cc}$;
3. $E = 1 - \sqrt{ee}$;
4. $c = 1 - \sqrt{CC}$;
5. $e = 1 - \sqrt{EE}$.

where *D*, *C*, *E*, *c*, *e* is the number of the gene-carrying persons in correlation with the number of the study subjects, *dd*, *cc*, *ee*, *CC* and *EE* is the corresponding phenotype frequency. The *Rh*-haplotypes frequency is computed by the formula proposed by A. E. Mourant:

1. $cde = \sqrt{ccddee}$;
2. $Cde = \frac{Ccddee}{2cde}$;
3. $cdE = \frac{ccddEe}{2cde}$;
4. $cDe = \frac{ccDee}{2cde}$;
5. $cDE = \sqrt{ccDEE + cdE^2}$ - cdE;
6. $CDe = \sqrt{CCDee + Cde^2}$ - Cde;
7. $CDE = \frac{CCDEe}{2(CDe + cde)}$,

where *ccddee*, *Ccddee*, *ccddEe*, *ccDee*, *CCDee* and *ccDEE* is the corresponding phenotypes' frequency.

The *RhD* and Kell-system alleles' concentration was computed under the following formula:

$$q = \sqrt{\frac{n_{aa}}{N}}, \quad p = 1 - q$$

where n_{aa} is the recessive homozygotes (*dd* and *kk*) by the indicated loci, *N* is the total number of the studied subjects.

In order to establish the concentration of the *MN*-system alleles, the following formulas were used:

$$P = \frac{n_A + \frac{1}{2}n_{AB}}{N}, \quad q = \frac{n_B + \frac{1}{2}n_{AB}}{N}$$

where n_A is the number of the *M*-phenotype carriers, n_{AB} - of the MN phenotype, and n_B is the number of the *N*-phenotype carriers.

The errors in the frequency of genes were computed by the formula: $M = \sqrt{P(100-P)/n}$ (28), where *P* is the frequency of antigens in %, *n* is the number of the study subject.

RESULTS

A whole number of peculiarities were registered in studying the Khulo region population for the erythrocytic group markers. 44±4.9% of the said region's population are carriers of the blood group 0(I), 37±4.82% - of the phenotypic group A(II). Rather higher is the ratio of the carriers of the B(III) blood group (17±3.7%). The AB(IV) blood group carriers constitute 2±1.4%

Studying concentrations of the ABO-system *r*, *p*, *q* alleles, it was revealed that the *r* allele concentration equaled 0.6. There is an insignificant difference between the concentrations of the *p*(0.21) and *q*(0.19) alleles (Fig. 1).

When studying individual alleles of the Rh system, the maximum frequency (100±0%) of the *e* antigens was revealed in the Khulo region population, followed by the frequency value of the *c* antigen (89±3.2%). The frequency of the *D*-antigen makes up 82±3.8%, while that of the *d*-antigen - 18±3.8% (Fig. 2). The *C*-antigen frequency is 61± 4.8%, and of the *E*-antigen - 23±4.2%.

The *e* alleles are characterized by the highest concentration of the Rh-system alleles in the Khulo region population. Their frequency in the said population equals 0.87; the concentration of the *c* allele is somewhat lower (0.64). The *D* allele concentration equals 0.58 and that of the *d* allele – 0.42. Relatively low is the *C* allele concentration, totalling 0.36. The lowest concentration value (0.13) is characteristic of the *E* allele.

When studying some genotypes of the Rh-system in Khulo region, high frequency distribution (77±4.2) of the *ee* version was identified. The number of the *Cc* genotype carriers was a little lesser (48±3.05). The *cc* genotype carriers' frequency made up 41±4.9%. As for the Ee genotype frequency, it equaled 23±4.2%. It is to be mentioned that the *EE* genotype is not generally represented in the Khulo region's population of the Ajarian Autonomous Republic (Fig. 3).

Six Rh-phenotypic groups with various frequency distributions were recorded for the Khulo region population. The most characteristic of the said region's population is the *CcDee* phenotype. Its frequency distribution totals 38±4.8%. Twice less frequency is characteristic of the *ccdee* (18±3.84%). The ratio of the *CcDEe* phenotype carriers totals 14±3.4%. The *CCDee*-phenotype frequency distribution equals 11±3.12%. Almost equal frequency is characteristic of the *ccDEe* (9±2.8%) and *ccDee* (10±3%) phenotypic groups (Fig. 6). In contrast to other regions of Ajara, the *Ccddee* and *ccDEE* phenotypic groups have not been registered in Khulo region.

Using statistical methods, only three *CDe*, *cDe*, *cde* haplotypes have been recorded for the Khulo region population. Other haplotypes are not generally met in the composition of the said region's population. The *CDe* haplotype is met with the 0.31 concentration. The concentration of the *cDe* haplotype equals 0.11. The highest frequency distribution is characteristic of the *cde* haplotype (0.42) (Fig. 5).

Six percent of the Khulo region population are carriers of the K factor and belong to the K(+) phenotypic group. As for the majority of the population, it is the K(-) phenotypic group carrier (Fig. 6).

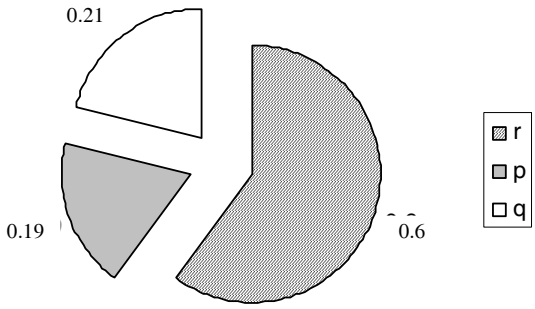

Fig. 1. Concentrations of the *ABO*-system alleles in the Khulo region population.

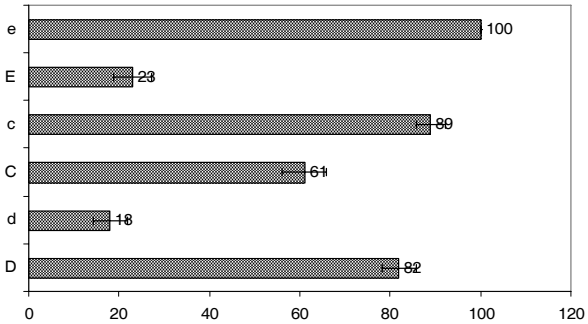

Fig. 2. Peculiarities of the Rh-system antigens distribution/frequency in the Khulo region population of Ajara.

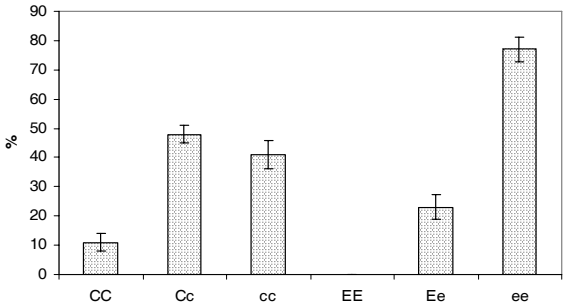

Fig. 3. Distribution peculiarities of the *CC, Cc, cc, Ee, EE, ee* genotypes in the Khulo region population of the Ajarian Autonomous Republic.

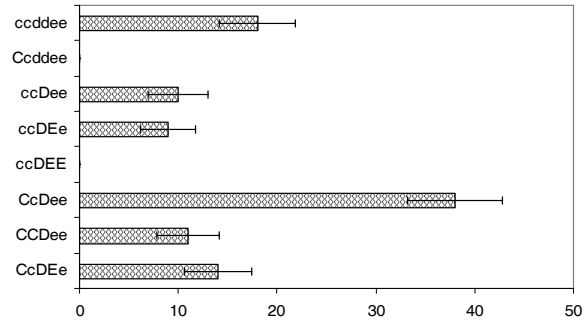

Fig. 4. Distribution peculiarities of the Rh-phenotypic groups in the Khulo region population of the Ajarian Autonomous Republic.

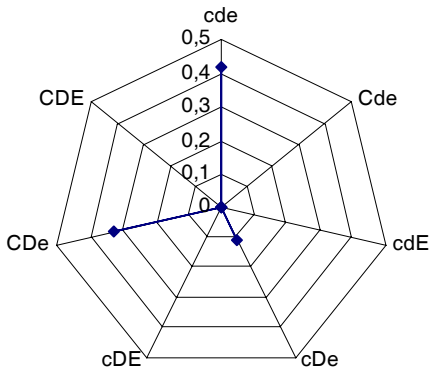

Fig. 5. Concentration of the *Rh* haplotypes in the Khulo region population.

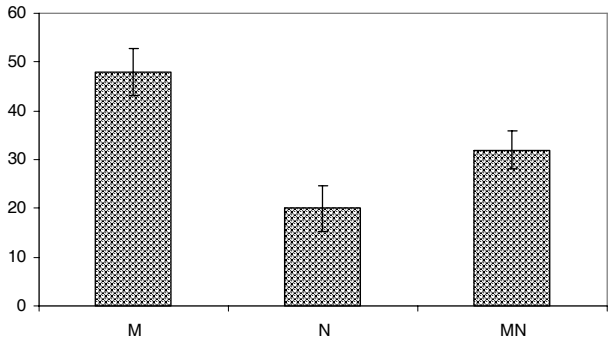

Fig. 7. Distribution peculiarities of the *MN*-system phenotypic groups in the Khulo region population of the Ajarian Autonomous Republic.

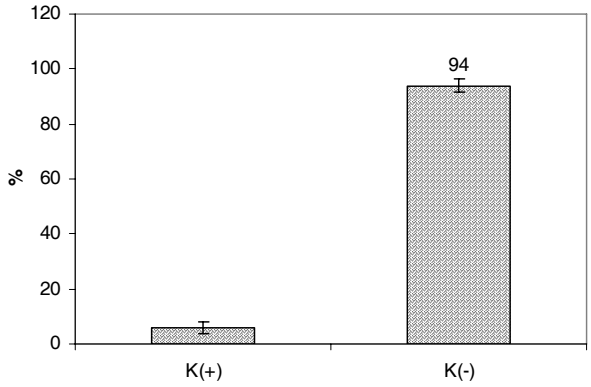

Fig. 6. Distribution peculiarities of the *Kell*-system phenotypic groups in the Khulo region population of the Ajarian Autonomous Republic.

Respectively, the concentration of K allele in the said population is minimal and equals 0.04, while that of k equals 0.96.

48±4.9% of the Khulo region population are carriers of the M-phenotypic group. The carriers of the MN-phenotypic group are 1.5-times less (32±4.6%). Of a relatively lower frequency (20±4%) is the N-phenotypic group (Fig. 7).

When studying the concentration of the MN-system alleles, a high concentration of the M allele was recorded for the Khulo region population. It equaled 0.58a. As for the *N*-allele concentration, it made up 0.42.

სამედიცინო მეცნიერებანი

ზოგიერთი იმუნოგენეტიკური მარკერების გავრცელების თავისებურებანი აჭარის მაღალმთიანი რაიონის (ხულო) მოსახლეობის მაგალითზე

მ. ნაგერვაძე, რ. ხუხუნაიშვილი, ა. დიასამიძე, მ. ქორიძე, გ. დუმბაძე, ლ. ახვლედიანი

შოთა რუსთაველის სახელმწიფო უნივერსიტეტი, ბათუმი

(წარმოდგენილია აკადემიკოს ფ. თოდუას მიერ)

სისხლის ერითროციტული ჯგუფური ანტიგენები გენეტიკურად მყარად დეტერმინირებულ თავისებურებას წარმოადგენს და მრავალმხრივი ბიოლოგიური და სამედიცინო მნიშვნელობა აქვს. ABO, Rh-Hr, Kell, MN

ერითროციტულ ჯგუფურ მარკერებზე გამოკვლეულ იქნა აჭარის ხულოს რაიონის მკვიდრი მოსახლეობა (105 კაცი). იმუნოსეროლოგიური მეთოდებით გამოკვლეულ იქნა სისხლში არსებული 13 ერითროციტური ჯგუფური ანტიგენი (ABO სისტემის ორი – A, B; Rh სისტემის შვიდი: C, C, C^w, D, D^u, E, e; Kell სისტემის ორი: K და k და MN სისტემის ორი: M და N ანტიგენი). მიღებული შედეგები დამუშავდა სტატისტიკურად. აღნიშნული რაიონის მოსახლეობის 44±4,9% აქვს 0(I) ჯგუფის სისხლი, 37±4,82% – A(II) ფენოტიპური ჯგუფის სისხლი. საკმაოდ მაღალია B(III) სისხლის ჯგუფის მატარებელთა ხვედრითი წილიც (17±3,7%). AB(IV) სისხლის ჯგუფი გვხდება 2±1,4% შემთხვევაში. Rh სისტემის ცალკეული ანტიგენების კვლევისას ხულოს რაიონის მოსახლეობაში გამოვლინდა e ანტიგენის მაქსიმალური სიხშირე (100±0%), მას რამდენადმე ჩამოუვარდება c ანტიგენის რაოდენობრივი მაჩვენებელი (89±3,12%). D ანტიგენი გვხდება 82±3,8% შემთხვევაში, d კი — 18±3,8% (სურათი 3). C ანტიგენის გავრცელების სიხშირე 61± 4,8%-ია, E ანტიგენისა კი — 23±4,2%. ხულოს რაიონის პოპულაციისათვის დაფიქსირებულ იქნა ექვსი Rh-ფენოტიპური ჯგუფი გარცელების სხვადასხვა სიხშირით. აღნიშნული რაიონის მოსახლეობისათვის ყველაზე მეტად დამახასიათებელია CcDee ფენოტიპი. მისი გავრცელების სიხშირე 38±4,8%-ია. მოსახლეობის უმრავლესობას K(-) ფენოტიპური ჯგუფის სისხლი აქვს, ხულოს რაიონის მოსახლეობის 48±4,9% კი M ფენოტიპური ჯგუფის მატარებელია. 1,5-ჯერ ნაკლებია MN ფენოტიპური ჯგუფის მქონეთა რაოდენობა (32±4,6%). შედარებით დაბალი სიხშირით (20±4%) გვხდება N ფენოტიპური ჯგუფი.

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