Journal of Biological Research **5**: 105 – 108, 2006
J. Biol. Res. is available online at http://www.jbr.gr

— SHORT COMMUNICATION —

Phenotype and gene frequencies of some blood groups and protein systems in Bulgarians from the Smolyan region (South-Central Bulgaria)

SOFIA D. BALTOVA

Department of Human Anatomy and Physiology, Faculty of Biology, University of Plovdiv "Paisij Hilendarski", 4000 Plovdiv, Bulgaria

Received: 28 August 2005 Accepted after revision: 27 January 2006

Studies on haemogenetic polymorphisms in Bulgarians are relatively scarce and mostly restricted to the Sofia region. As a contribution to the haemogenetic characteristics of Bulgarians, the results of an examination of ABO, Rh, MNSs, P1, HPA, GM1, PGM1, ACP and ESD in 540 individuals from the Smolyan region of south-central Bulgaria are reported. The typing was done by standard techniques. During all surveys, only the classic types of blood group systems were found. The combined data are compared with other populations.

Key words: blood group, phenotype, genotype, enzyme, polymorphism.

INTRODUCTION

The survey of blood system polymorphism gives answers to some general biological problems such as the microevolutionary changes taking place constantly in contemporary humans in the process of their interaction with the conditions of the environment in which they live. The stability of blood group factors as well as their determination by means of objective and relatively simple methods makes them particularly convenient for population surveys and provides an opportunity to identify mutations and phenomena like isolation, migration and genetic drift.

The various populations are typical with their own aggregates of features that can be predetermined by mixing or isolation, genetic mutations or chromosome combinations, or adaptation capacities to different ecological situations.

Studies on haemogenetic polymorphisms in Bulgarians are relatively scarce and mostly restricted to the Sofia region (e.g. Karamihova-Tsacheva, 1967; Boev & Popwassilew, 1969; Ananthakrishnan & Walter, 1972; Rupcheva, 1972; Kalchev, 1980). We still have insufficient data on phenotype and genotype distributions for other towns and regions.

The purpose of the present survey was to determine the phenotype distribution and gene frequencies of blood group systems aiming at the clarification of the genetic status of the population from the district of Smolyan, which constitutes a part of the south-central region of Bulgaria. Then, comparisons with other populations will follow. To our knowledge, there are no surveys on serum and enzyme systems in this region.

MATERIALS AND METHODS

We studied 540 individuals of both sexes, aged between 18 and 45 years from the Smolyan region of south-central Bulgaria. They were clinically healthy and were unrelated to each other.

For the study of the blood group (ABO, Rh, MNSs, P1, serum), HPA, GM1 and enzyme systems (phosphoglucomutase, PGM1, E.C. 2.7.5.1.; acid phosphatase, ACP, E.C. 3.1.3.2.; esterase D, ESD, E.C. 3.1.1.1.), parallel investigations were made. The

Corresponding author: tel.: +359 / 032 629 098, e-mail: bsofi@abv.bg

typing was done by standard techniques.

The statistical analysis was performed using the methods of alternative, correlation and non-parametric analyses (Lakin, 1990). The principal component analyses were done with the SPSS program.

RESULTS AND DISCUSSION

During all surveys, only the classic types of blood group systems were found. In the surveyed contingent, the majority belonged to the phenotypes in erythrocyte systems (A_1: 43.33%, CcDee: 33.33%, MNS: 34.44%, $P1^+$: 56.67%), in serum systems (HPA 2-2: 58.89%, $GM1^-$: 66.67%) and in enzyme systems (ACP B: 37.78%, ESD1: 85.56%, PGM 2-1: 54.44%). The other most rarely found versions were: A_2: 2.22%, Ccddee and CCDEe: 1.11%, NS: 3.33%, P^-: 43.33%, HPA 1-1: 7.78%, $GM1^+$: 33.33%, ACP A: 4.44%, PGM 2-2: 12.22%, ESD 2-2: 2.22%.

The surveyed contingent of persons without any kinds of relations among them provided an opportunity to calculate the incident gene frequencies (Table 1).

By using the Pearson's criterion, comparison of the distribution between the monitored and expected values yielded an insignificant difference ($p > 0.05$). In some cases, a test for Hardy-Weinberg equilibrium was impossible due to missing degrees of freedom, but mostly the observed frequencies corresponded well to the expected values.

The present survey constitutes a continuation of our previous studies on blood group systems among Bulgarian populations from the south-central and south-eastern regions of the country where we made comparisons with data from other Balkan populations. That survey shows a single cluster with southern Romania and Hungary, while the other populations are more separated from this cluster and from each other (Baltova *et al*., 2005). Again, the genetic distances are relatively small. The data for the non-Bulgarian populations (Albania, Greece, FYROM, Romania, Hungary) were taken from the literature (Czeizel *et al*., 1991; Schmidt *et al*., 2000; Scheil *et al*., 2001).

Comparisons of the results of the tests with those concerning the Bulgarian populations described by other authors and from data in the available literature were also made.

Regarding the role of the genetic markers, the results of investigations on blood group systems (ABO, Rh, MNSs, P1) (Ilieva, 1956; Stojanov, 1959; Popwassilew & Rackwitz, 1962; Zographov, 1962; Boev & Popwassilew, 1969), on gene frequencies of HPA and GM1 (Popwassilew, 1962; Karamihova-Tsacheva, 1967; Walter *et al*., 1972) and enzyme (PGM1, ESD, ACP) polymorphisms (Ananthakrishnan & Walter, 1972; Rupcheva, 1972; Kalchev, 1980; Peev, 1980) should be emphasized.

The phenotype and gene frequencies of blood group systems vary in the different populations and on this basis, their serological characteristics are formed. The informative value of a given system depends on the intensity of the existing geographical, racial and national differences in the frequencies of its component alleles. None of them showed an identical variability.

From the literature data it is evident that among the persons tested in the district of Smolyan, the gene frequencies in the separate blood group systems are slightly increased for the alleles A_1, B, MS, Ms, NS, cDe, Cde, ACP^B, PGM^2, and somehow higher for the cde, cDE, CDE, $P1^-$, ACP^C, ESD^1, HPA^2, and $GM1^-$. For the alleles A_2, O, Ns, $PGM1^1$ they are slightly decreased, while in the $P1^+$, ACP^A, ESD^2, HPA^1 and $GM1^+$, the decrease is greater.

The lower values for the frequency of the alleles in the surveyed group of Bulgarians correspond to the historical and geographical data related to their origin. This population has not yet experienced the migration processes and the people who remained to live there are of native origin. The genetic memory of previous populations has been preserved in them to a certain extent. The potential for genetic change exists mostly within the population unit and the rate of change is dependent on the effect of drift.

The values of the allele frequency of different Bulgarian subpopulations vary within narrow limits with a certain tendency of increasing from the south to the north and from the west to the east.

The results of our surveys as a whole also comply with those on other European populations (Mourant *et al*., 1976; Pap, 2000; Scheil & Huckenbeek, 2000; Schmidt *et al*., 2000; Scheil *et al*., 2001; Schmidt & Scheil, 2001; 2003). According to the gene frequencies, the Bulgarian population examined here belongs to the broader European population, which is also indicated by other blood group, serum and enzyme systems.

TABLE 1. The distribution of phenotypes and gene frequencies in blood groups, serum and enzyme systems from the Smolyan region

Phenotype	**N = 540**				
	Obs.	**%**	**Exp.**	**llele and genotype frequencies**	
A_1	234	43.33	218.64	A1	0.2812
A_2	12	2.22	9.96	A2	0.0161
B	114	21.11	95.28	B	0.1394
0	156	28.89	171.36	0	0.5633
A_1B	24	4.45	42.06		
A_2B	–	–	2.70		
			df = 0		
ccddee	72	13.33	58.89	cde	0.3295
ccDee	36	6.67	29.62	cDe	0.0741
ccDEe	24	4.44	61.65	Cde	0.0164
ccDEE	12	2.22	11.03	Cde	0.4245
Ccddee	6	1.11	6.09	cDE	0.1409
CcDee	180	33.33	186.57	CDE	0.0147
CcDEe	114	21.11	73.77		
CCDee	90	16.68	105.09		
CCDEe	6	1.11	7.29		
			df = 0		
MS	138	25.56	151.08	MS	0.3108
Ms	54	10.00	46.92	Ms	0.2948
MNS	186	34.44	159.06	NS	0.0837
MNs	84	15.56	98.94	Ns	0.3107
NS	18	3.33	31.86		
Ns	60	11.11	52.14		
			$\chi^2 = 2.7094$, df = 2, $20 < p < 30$		
P1+	306	56.67	–	P1+	0.3417
P1-	234	43.33	–	P1-	0.6583
HPA 1-1	42	7.78	32.28	HPA^1	0.2444
HPA 2-1	180	33.33	199.44	HPA^2	0.7556
HPA 2-2	318	58.89	308.28		
			$\chi^2 = 0.8547$, df = 1, $30 < p < 50$		
GM1+	180	33.33	–	GM1	0.1835
GM1-	360	66.67	–	nonGM1	0.8165
ACP A	24	4.44	28.02	ACP*A	0.2278
ACP B	204	37.78	212.82	ACP*B	0.6278
ACP C	–	–	11.28	ACP*C	0.1444
ACP AB	156	28.89	154.44		
ACP AC	42	7.78	35.52		
ACP BC	114	21.11	97.92		
			$\chi^2 = 2.7495$, df = 2, $20 < p < 30$		
ESD 1	462	85.56	453.78	ESD*1	0.9167
ESD 2-1	66	12.22	82.50	ESD*2	0.0833
ESD 2	12	2.22	3.72		
			df = 0		
PGM1 1	180	33.34	198.06	PGM1*1	0.6056
PGM1 2-1	294	54.44	257.94	PGM1*2	0.3944
PGM1 2	66	12.22	84.00		
			$\chi^2 = 1.7576$, df = 1, $10 < p < 20$		

"df = 0" = no degrees of freedom for Hardy-Weinberg testing

CONCLUSIONS

According to the allozyme analysis, the most frequent are the homozygotes. The allele frequencies of the ACP, ESD and PGM systems are as follows: ACP^A: 0.2278, ACP^B: 0.6278, ACP^C: 0.1444; ESD^1: 0.9167, $ESTD^2$: 0.0833; PGM^1: 0.6056, PGM^2: 0.3944.

The variation of the blood group, serum and enzyme systems in the studied Bulgarian population from Smolyan does not differ substantially from that of the remaining Bulgarians.

REFERENCES

Ananthakrishnan R, Walter H, 1972. Red cell enzyme polymorphisms in Bulgaria. *Humangenetik*, 15: 186-190.

Baltova S, Scheil H-G, Schmidt HD, 2005. Haemogenetic studies in Bulgaria. *Anthropologischer anzeiger*, 63: 141-151.

Boev P, Popwassilew I, 1969. Zur Häufigkeit der Blutgruppen ABO, MN und P sowie der Serumgruppen Hp und Gm in Bulgarien. *Anthropologischer anzeiger*, 31: 184-188.

Czeizel H, Benkmann H-G, Goedde HW, 1991. *Genetics of the Hungarian population*. Springer Verlag, Berlin.

Ilieva V, 1956. Studies on the spread of erythrocyte antigens M and N among Bulgarians. *Comptes rendus de l' academie bulgare des sciences*, 9: 69-71.

Kalchev Y, 1980. PGM in Bulgaria. Population genetics and forensic medicine research. Ph. D. Thesis, Sofia (in Bulgarian).

Karamihova-Tsacheva L, 1967. Distribution of the haptoglobin types in Bulgaria. *Comptes rendus de l' academie bulgare des sciences*, 20: 637-640.

Lakin G, 1990. *Biometria*. Vishaya shkola, Moskva (in Russian).

Mourant AE, Kopeć AC, Domaniewska-Sobczak K, 1976. *The distribution of the human blood groups and other polymorphisms* (2nd edition). Oxford University Press, London.

Pap M, 2000. Population genetic research in Hungary. *Acta biologica szegediensis*, 44: 129-133.

Peev Hr, 1980. Polymorphism of enzyme EsD in inhabitants from South Bulgaria. Ph. D. Thesis, Plovdiv (in Bulgarian).

Popwassilew I, 1962. Über die Verteilung der Haptoglobintypen in Sofia. *Zeitschrift für ärztliche fortbildung*, 56: 770.

Popwassilew I, Rackwitz A, 1962. Über die Häufigkeit des Faktors P1 (im allgemeinen als P-positiv bezeichnet) in Bulgarien. *Zeitschrift für ärztliche fortbildung*, 56: 764.

Rupcheva L, 1972. Polymorphism of ACP of erythrocyte. Ph. D. Thesis, Sofia (in Bulgarian).

Scheil H-G, Huckenbeek W, 2000. Hamogenetische Studien im Regierungsbezirk Köln (Nordrhein-Westfalen, Deutschland). *Anthropologischer anzeiger*, 58: 155-169.

Scheil H-G, Scheffrahn W, Schmidt HD, Huckenbeck W, Efremovska L, Xirotiris N, 2001. Population genetic studies in the Balkans. I. Serum proteins. *Anthropologischer anzeiger*, 59: 203-211.

Schmidt HD, Scheil H-G, Scheffrahn W, 2000. The history and genetics of the Aromun population. In: Susanne C, Bodzsar E, eds. *Human population genetics in Europe*, Vol. 1. Biennial Books of EAA, Eotvos University Press, Budapest: 29-37.

Schmidt HD, Scheil H-G, 2001. Untersuchungen über die Variabilität der Blutgruppenhäufigkeiten in Rumanien. *Anthropologischer anzeiger*, 59: 227-232.

Schmidt HD, Scheil H-G, 2003. Blood group frequencies in Romania: Microregional and ethnic differencies. *Anthropologischer anzeiger*, 61: 381-393.

Stojanov S, 1959. Zur Frage der Rho – Antigenverbreitung in Bulgarien. *Zentralblatt der gynäkologie*, 81: 797-799.

Walter H, Ananthakrishnan R, Tsacheva L, 1972. Serum protein groups in Bulgaria. *Human heredity*, 22: 529-532.

Zographov DG, 1962. ABO-Blutgruppen und bösartige Geschwülste. *Oncologia*, 15: 59-64.