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Properdin Factor B (Bf) Polymorphism in the Population of Veneto, Italy*,**

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Summary. The distribution of Bf phenotypes in the population of Veneto was investigated by agarose gel electrophoresis and immunofixation. In our sample ($n = 592$), the seven common phenotypes F, S, F-S, S-S0.7, S-F1, F-S0.7, F-F1 were observed and the following gene frequencies calculated: Bf*S = 0.7399; Bf*F = 0.2280; Bf*F1 = 0.0177; Bf*S0.7 = 0.0144. These gene frequencies are compared to those found in other populations. Analysis of 21 mother-child pairs was in agreement with an autosomal codominant inheritance.

Key words: Properdin factor B, genetic polymorphism (Bf) in the Veneto population – Blood groups, properdin factor B

Zusammenfassung. Die Verteilung der Bf-Phänotypen wurde bei der venetischen Bevölkerung mittels Agarosegel-Elektrophorese und Immunofixation untersucht. In unseren Stichproben wurden die Bf-Phänotypen F, S, F-S, S-S0,7, S-F1, F-S0,7, F-F1 beobachtet und folgende Genfrequenzen berechnet: Bf*S = 0,7399; Bf*F = 0,2280; Bf*F1 = 0,0177; Bf*S0,7 = 0,0144. Diese Genfrequenzen wurden mit den bei anderen Bevölkerungsgruppen gefundenen Frequenzen verglichen. Die Untersuchung von 21 Mutter-Kind-Paaren zeigte eine autosomal-kodominante Relation.

Schlüsselwörter: Properdinfaktor B (Bf), genetischer Polymorphismus (Bf) der venetischen Bevölkerung – Blutgruppen, Properdinfaktor B

Introduction

Properdin factor B (Bf), also known under the designation glycine-rich-B-glycoprotein (GBG) or C3 proactivator, shows a polymorphism discovered in 1972

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by Alper et al. [1]. This marker is the product of two common codominant alleles designated in the nomenclature of Mauff et al. [2] as Bf*S and Bf*F and two less common Bf*S0.7 and Bf*F1.

A number of alleles with low gene frequencies are also found and classified according to the relative mobilities of the gene product [2]. Furthermore, the existence of a silent allele Bf*Q0 was reported [3–6].

In this paper we report the distribution of Bf phenotypes and the gene frequencies in the Veneto population; they are compared with those of other authors.

Materials and Methods

Sera from 592 unrelated apparently healthy blood donors were provided by the Transfusion Centre of the Civil Hospital of Padua. Serum samples from mother-child pairs were obtained from the routine case material of our institute. Determination of Bf phenotypes was carried out by high voltage agarose gel electrophoresis (HVAGE) followed by immunofixation according to Dykes [7–8], with minor modifications, as described above.

The chamber buffer was a barbital buffer, pH 8.6, for the gel this was used diluted 1:3 with distilled water. The agarose gel (1%) was 1.25 mm thick. Electrophoresis was carried out on a plate sized 20 × 20 cm, an efficient cooling system was needed.

Undiluted serum samples were applied at 2 cm distance from the cathode end and submitted to electrophoresis at 20 V/cm with a separation time of 3 h. For immunofixation we used anti-human properdin factor B serum from ATAB 1:4 diluted in saline. Staining was performed with Coomassie blue R250.

Results and Discussion

The pattern of bands results from Bf typing on HVAGE as showed in Fig. 1. For the classification of the samples we have adopted the nomenclature according to Mauff [2], based on the relative electrophoretic mobilities of all phenotypes which are not common S or F, using the distance S-F1 as reference.

The distribution of Bf phenotypes and the gene frequencies are shown in Table 1.

In a total of 592 samples, four of the possible Bf alleles in white people were observed.

Our results for Bf phenotypes provide a satisfactory fit to the Hardy-Weinberg equilibrium ($0.90 < P < 0.95$ at 3 df).

Our results were compared to those obtained in other Italian regions and in other countries. All these gene frequencies are reported in Table 2.

The Bf allele frequencies found in Veneto are similar to those of other Italian regions and all white populations excluding Sardinia where the Bf*F1 allele shows a higher frequency, but this concerns a geographically isolated population.

Relatively to the results obtained from Minnesota Whites, Eskimo, Amerindians and US Blacks like those observed by Dykes [17], we can observe three groups in which the white people represented the group characterized by a high frequency of Bf*S and Bf*F. The others are represented by Eskimos and

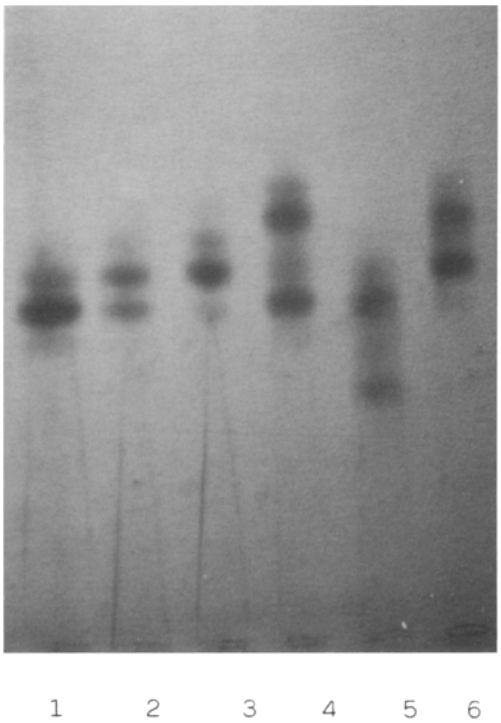

Fig. 1. Bf banding patterns after high voltage agarose gel and immunofixation. From *left* to *right:* 1(S), 2(F/S), 3(F), 4(S/F1), 5(S/S0.7), 6(F/F1)

Table 1. The distribution of Bf phenotypes and the Bf gene frequencies in the Veneto population

Bf phenotypes	No. observed	Observed (%)	No. expected
S	324	54.7297	324.0565
FS	199	33.6149	199.7616
F	31	5.2365	30.7863
S-S0.7	13	2.1959	12.6792
S-F1	16	2.7027	15.6401
F-S0.7	4	0.6757	3.9781
F-F1	5	0.8446	4.7898
S0.7	–	–	0.1221
F1	–	–	0.1863
	592	100.0000	592.0000

Gene frequencies: Bf*S = 0.7399; Bf*F = 0.2280;
Bf*S0.7 = 0.0144; Bf*F1 = 0.0177

$\chi^2 = 0.4104$; for 3 *df:* $0.90 < P < 0.95$

Table 2. Comparison of Bf allele frequencies in several populations

No. of cases	Populations (authors)	Bf*S	Bf*F	Bf*S0.7	Bf*F1	Others
592	Veneto (this study)	0.7399	0.2280	0.0144	0.0177	–
1000	Tuscany [9]	0.7130	0.2495	0.0254	0.0130	–
191	North-West Italy [10]	0.746	0.237	0.010	0.007	–
128	North-East Italy [10]	0.785	0.180	0.016	0.019	–
123	Central-Italy [10]	0.679	0.280	0.033	0.08	–
165	South-Italy [10]	0.715	0.258	0.018	0.09	–
217	Sardinia [11]	0.5783	0.2189	0.0046	0.1982	–
1660	Sweden [12]	0.8139	0.1735	0.0042	0.0084	–
300	Norway [13]	0.817	0.172	0.007	0.005	–
654	Switzerland [14]	0.805	0.176	0.009	0.001	0.0004
1245	Western Germany [15]	0.8084	0.1743	0.0092	0.0077	0.0004
522	Germany (Hessen) [16]	0.7998	0.1772	0.0163	0.0077	–
1005	Minnesota Whites [8]	0.7985	0.1870	0.0040	0.0105	–
368	Eskimos [17]	0.9891	0.0090	–	0.0013	–
510	Amerindians (Apache) [17]	0.9862	0.0039	0.0049	–	0.0049
357	US Blacks [17]	0.4475	0.5014	0.0028	0.0182	–

Table 3. Distribution of Bf phenotypes in 21 mother-child pairs

Mothers		Children			
		S	FS	F	
S	14	12	2	–	14
FS	7	2	5	–	7
F	–	–	–	–	
	21				21

Amerindians with a total absence of Bf*F1 allele and Blacks with an equal frequency of Bf*F and Bf*S.

Finally, 21 mother-child pairs were investigated (Table 3). The segregation of Bf phenotypes was in accordance with the genetic model of an autosomal codominant mode of inheritance.

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