

THE POLYMORPHISM OF Bf AND C3 IN THE POPULATION OF VENETO (ITALY)

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INTRODUCTION

A genetic polymorphism of human C3 was demonstrated by Wieme and Demeulene² in 1967 (25) and later, confirmed simultaneously by Alper and Propp, Azen and Smithies (2, 5). They used electrophoretic techniques either in agarose gel or starch gel. They described a system with two codominant alleles C3^S and C3^F. Further studies revealed 22 less common variants which have been designated according to their electrophoretic mobility. Bf also shows a polymorphism discovered by Alper et al. in 1972 (1) and then a number of rare alleles have been described. The structural gene locus Bf is characterized by some alleles of which are known two common F and S, two less common F1 and S0.7 and some others less frequent in according to their electrophoretic relative mobilities(13). Both these complement components also been examined from two points of view: the population genetics and the medico-legal application. The purpose of this paper is to present the results of a study of the distribution of C3 and Bf phenotypes in the Veneto population.

MATERIAL AND METHODS

Serum samples, 740 for C3 and 592 for Bf ,were obtained from healthy blood donors from Padua Hospital Blood Bank (Veneto Italy). Samples were stored at -30°C prior to analysis and typed within 2-3 days. High voltage gel electrophoresis , as published by Tiesberg (23) and modified by Domenici et al. (11) was used for phenotyping the C3 component. It consisted of a continuous buffer system with barbital/ calcium lactate buffer. Electrophoresis was carried out on glass plate covered with 1% agarose gel. Serum samples diluted in saline, were applied cathodally. At the end of

the run, each plate was fixed in 5% TCA, dried, and finally, stained with CBB-R250.

Determination of Bf phenotypes was carried out by high voltage agarose gel electrophoresis followed by immunofixation according to Dykes(12,13). Electrophoresis and gel buffer was a barbital buffer pH 8.6. Undiluted serum samples were electrophoresed at 20 V/cm for 3 hours; an efficient cooling system was needed.

For immunofixation a cellulose acetate strip, saturated with Anti-human Properdin Factor B serum was placed for at least 60 min on the gel.

The staining was performed with Coomassie Blue R250.

RESULTS AND DISCUSSION

Table 1 illustrates the observed and expected distribution of phenotypes and gene frequencies of 740 blood donors of Veneto population tested for C3. The population is in Hardy-Weinberg equilibrium with a chi-square=0.048 $0.95 < p < 0.98$ for 1 d.f.

In table 2 our gene frequencies are compared to those found in other populations with similar studies: there is good accordance with the results obtained by other Authors in European countries. The most common gene in the populations studied up to now is C3^S.

In Table 3 the distribution of Bf phenotypes and of Bf alleles in a sample of 592 unrelated individuals from Veneto region is shown. A good fit to the Hardy-Weinberg law was observed.

In Table 4 our gene frequencies are compared with other found in some Caucasian populations using the same technique of electrophoresis followed by immunofixation.

The most common gene in the population studied at present is Bf^S characterized by a frequency ranging from 0.5783 (Sardinia) to 0.8084(Germany). Bf^{S0.7} and Bf^{F1} show similar frequencies, always lower than Bf^F;

The C3 and Bf systems are informative, the methods for their determination are simply-reproducible and of relatively low cost, so that they appear useful for both population studies and cases of disputed paternity.

TABLE 1. C3 distribution in Veneto

PHENOTYPES	OBSERVED		EXPECTED	
	n	%	n	%
S	482	0.65135	482.43798	0.65194
FS	230	0.31081	229.30790	0.30988
F	27	0.03649	27.24817	0.03682
S-VF F-VF VF	1	0.00135	1.00595	0.00136
	740	1.00000	740	1.00000

$C3^S=0.80743$, $C3^F= 0.19189$, $C3^V=0.00068$
 $\chi^2=0.0048$.95 < p < .98 for 1df

TABLE 2. Comparison of C3 allele frequencies in several Populations

No. of cases	Populations (Authors)	$C3^S$	$C3^F$	$C3^V$
740	Veneto(this study)	0.80743	0.19189	0.00068
650	Tuscany(Domenici 1985)	0.803	0.195	0.001
325	Rome(Scacchi 1979)	0.791	0.203	0.006
950	Sicily(Bonavita 1985)	0.815	0.184	-
1034	Finland(Arvilommi 1973)	0.829	0.170	-
2454	Norway(Teisberg 1970)	0.786	0.208	0.005
213	Sweden(Bronnestam 1971)	0.770	0.230	-
406	Demark(Dissing 1971)	0.816	0.182	0.001
2340	Germany(Rittner 1974)	0.787	0.202	0.010
1988	Germany(Argawall 1972)	0.784	0.215	0.004
1326	Germany(Geserik 1980)	0.813	0.183	0.003
889	Switzerland(Pflugshupt 1973)	0.773	0.221	0.006
818	Belgium(Hoste 1977)	0.811	0.186	0.003
961	Spain(Argawall 1972)	0.782	0.211	0.006
388	China(Zhao 1983)	0.996	0.003	-
29	American Negroes (Alper 1978)	0.90	0.10	-

TABLE 3. The distributio of Bf phenotypes and gene frequencies in the Veneto population.

PHENOTYPES	OBSERVED		EXPECTED	
	n	%	n	%
S	324	54.72973	324.05653	54.73928
FS	199	33.61486	199.76163	33.74352
F	31	5.230649	30.78630	5.20039
S-SO.7	13	2.19595	12.67917	2.14175
S-F1	16	2.70270	15.64012	2.64191
F-SO.7	4	0.67568	3.97813	0.67198
F-F1	5	0.84459	4.78975	0.80908
SO.7	-	-	0.12207	0.02062
F1	-	-	0.18630	0.03147
	592	100.00000	592.00000	100.00000

$$Bf^S=0.73986, Bf^F=0.22802, Bf^{SO.7}=0.01436, Bf^{F1}=0.01774$$

$$\chi^2=0.3385 \quad .95 < p < .98 \quad \text{for } 3 \text{ df}$$

TABLE 4. Distribution of Bf phenitypesin some Caucasian populations (typed by HVAGE and immunofixation)

No. of cases	Population (Authors)	Bf ^S	Bf ^F	Bf ^{SO.7}	Bf ^{F1}	other
592	Veneto(this study)	0.73986	0.22804	0.01436	0.01774	-
1000	Tuscany(Domenici 1985)	0.7130	0.2495	0.0254	0.0130	-
400	Sicily(Crinò 1985)	0.8063	0.1688	0.0212	0.0025	0.0012
191	North-West.Italy	0.746	0.237	0.010	0.007	-
128	North-East.Italy	0.785	0.180	0.016	0.019	-
123	Central Italy	0.679	0.280	0.033	0.008	-
165	Southern Italy	0.715	0.258	0.018	0.009	-
217	Sardinia(Davrinche 1984)	0.5783	0.2189	0.0046	0.1982	-
1660	Sweden(Hjalmarsson 1981)	0.8139	0.1735	0.0042	0.0084	-
300	Norway(Teisberg 1977)	0.817	0.172	0.007	0.005	-
1245	Western-Germany(Mauff 1975)	0.8084	0.1743	0.0092	0.0077	0.0004
522	Germany-Hessen(Kühnl 1978)	0.7998	0.1772	0.0163	0.0077	-
1005	Minnesota Whites (Dykes 1980)	0.7985	0.1870	0.0040	0.0105	-

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