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GENETIC POLYMORPHISM OF PROPERDIN FACTOR B (BF) IN A CHINESE POPULATION: EXISTENCE OF TWO RARE VARIANTS

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Summary Genetic polymorphism of properdin factor B (BF) was investigated in sera from 259 healthy individuals living in Guangzhou, southern China, by means of high voltage agarose gel electrophoresis followed by immunofixation. Besides the common phenotypes, SS, FS, FF and SS07, two rare heterozygous phenotypes tentatively named as SSG1 and SFG2 were observed. The allele frequencies estimated for *BF*S*, *BF*F*, *BF*S07*, *BF*SG1* and *BF*FG2* were 0.8668, 0.1197, 0.0077, 0.0019 and 0.0039, respectively. Family data indicated an autosomal, codominant inheritance for the *BF*FG2* allele.

INTRODUCTION

Properdin factor B (BF) is a proactivator in the alternative pathway of complement activation. Genetic polymorphism of human BF was first described by Alper *et al.* (1972). The interest in the BF polymorphism has been increased since the BF locus was reported to be linked to the HLA loci on chromosome 6 (Allen, 1974; Rittner *et al.*, 1975; Olaisen *et al.*, 1975). At least 20 BF allotypes have been described so far (Mauff *et al.*, 1978; Larsen *et al.*, 1981; Davrinche *et al.*, 1982; Tokunaga *et al.*, 1982; Dykes *et al.*, 1981, 1983). Considerable geographical differences were noted in the distribution of BF allotypes.

In the mainland Chinese populations, there was only a single report of BF polymorphism (Zhao, 1983). It dealt with a population from Shanghai, east-central China, and described the frequences for three common alleles, *BF*F*, *BF*S* and *BF*S07*. Recently, we examined the BF types among patients of diabetes mellitus in Guangzhou, southern China (Luo *et al.*, 1986). In this paper, we describe the detailed data on the BF polymorphism in healthy adults living in Guangzhou city.

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MATERIALS AND METHODS

Blood samples were obtained from 259 unrelated healthy adults living in Guangzhou city who attended routine antenatal clinic for genetic counselling at the First Affiliated Hospital of Sun Yat-sen University of Medical Sciences. Specimens for family study were obtained from a family of a propositus showing a rare BF variant. BF typing was carried out by means of a high voltage agarose gel electrophoresis and subsequent immunofixation according to Alper *et al.* (1972), with slight modifications (Tokunaga *et al.*, 1982).

RESULTS AND DISCUSSION

The results of BF typing in the present study are shown in Table 1. In agreement with the result of Zhao (1983) on east-central China, the present study showed the occurrence of three common BF alleles, *BF*S*, *BF*F* and *BF*S07*, and the absence of *BF*F1*.

Table 2 lists the frequencies of the common BF alleles in various populations thus far reported. The frequency of *BF*S* is higher than that of *BF*F* in the present study, corresponding to the other reports on Mongoloid and Caucasoid populations. The distribution of BF allele frequencies in Guangzhou population was found to be similar to that in the populations from east-central China, and probably also from Thailand and in Lapps, but considerably different from those in Japanese and Koreans. These latter populations appears to have slightly higher frequencies for *BF*F* than in Chinese populations, and lack *BF*S07*. Marked deviation in the gene frequency distribution is notable between Asian and African populations.

Besides the common BF phenotypes, two unusual types were identified, each of which was considered to be heterozygous for a rare variant BF allele (Fig. 1). One of these denoted as type SSG1 was found in a single individual, and showed a BF

Table 1. Distribution of phenotypes and allele frequencies of BF in southern Chinese.

Phenotypes	N. Obs. (%)	N. Ex. (%)	Allele frequency
SS	197 (76.06)	194.60 (75.14)	*BF*S*: 0.8668±0.0149
FS	48 (18.53)	53.75 (20.75)	*BF*F*: 0.1197±0.0143
FF	7 (2.70)	3.71 (1.43)	*BF*S07*: 0.0077±0.0035
SS07	4 (1.50)	3.46 (1.34)	*BF*SG1*: 0.0019±0.0017
SSG1	1 (0.39)	0.85 (0.33)	*BF*FG2*: 0.0039±0.0025
SFG2	2 (0.77)	1.75 (0.68)	
Total	259(100.00)	258.12 (99.66)	1.0000

$\chi^2=2.476$, 1 d.f., $p>0.10$.

Table 2. Comparison of BF allele frequencies in different populations.

Population	N.ex.	BF allele frequency				*P*	Authors
		S	*F*	*F1*	*S07*		
S. Chinese	259	.867	.120	0	.008	—	This study
E. Chinese	200	.870	.127	0	.003	ns	(1)
Thai	184	.902	.098	0	0	ns	(2)
Korean	220	.775	.225	0	0	**	(3)
Japanese	487	.801	.198	0	0	**	(4)
Saudi Arabian	246	.474	.447	.020	.059	**	(5)
North African (Negroid)	944	.282	.655	.034	.025	**	(6)
Cameroon	275	.373	.595	.029	.003	**	(5)
Tunisian	375	.617	.281	.019	.083	**	(7)
Mali	457	.300	.640	.046	.014	**	(7)
Spaniard	330	.658	.266	.052	.022	**	(8)
French (Basque)	201	.550	.296	.139	.015	**	(9)
Dane	318	.780	.203	.008	.009	**	(10)
West German	1,245	.808	.174	.008	.009	*	(6)
Norwegian	300	.817	.172	.005	.007	*	(11)
Lapp (Norway)	197	.888	.112	0	0	ns	(11)
North American (Caucasoid)	158	.709	.278	0	.013	**	(12)
North American (Negroid)	127	.437	.512	.051	0	**	(12)

P denotes the probability for a chi-square heterogeneity test between data of this study and those of other reports: ns, not significant, * significant at 1% level, ** significant at 0.1% level. Authors: (1) Zhao, 1983, (2) Greiner *et al.*, 1980, (3) Park *et al.*, 1985, (4) Tokunaga *et al.*, 1982, (5) Goedde *et al.*, 1979a, 1979b; (6) Mauff *et al.*, 1975; (7) Davrinche *et al.*, 1981; (8) Rodriguez-Córdoba *et al.*, 1981; (9) Ohayon *et al.*, 1980; (10) Mortensen and Lamm, 1981; (11) Teisberg and Olaisen, 1977; (12) Alper *et al.*, 1972.

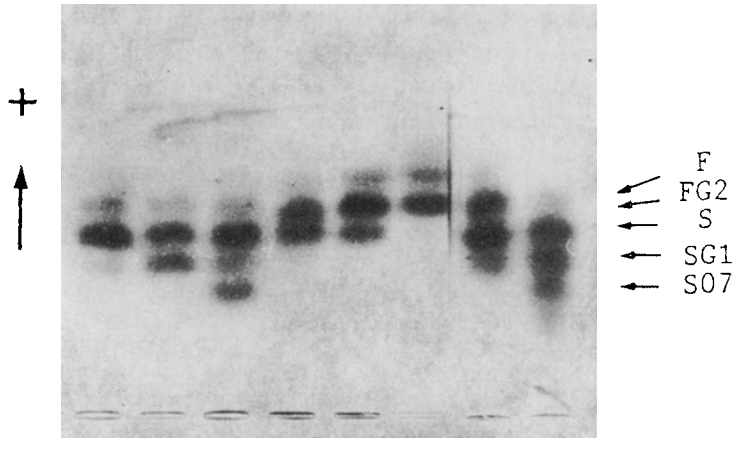

Fig. 1. Photograph of BF phenotypes observed in southern Chinese. 1, SS; 2, SSG1; 3, SS07; 4, SFG2; 5, FS; 6, FF; 7, FS+SSG1 (mixture); 8, SS07+SSG1 (mixture).

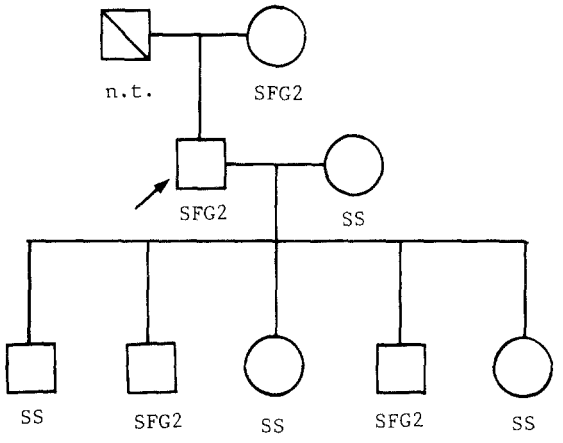

Fig. 2. A pedigree suggesting the autosomal, codominant inheritance of *BF*FG2*. n.t., not tested. The arrow indicates the propositus.

band intermediate between S and S07 corresponding the position of the cathodal minor band of type SS. When the plasma was mixed with those of types FS and SS07, respectively, three-band patterns emerged (Fig. 1, lane 7 and 8). The relative mobility of this variant was estimated to be within the range of 0.35–0.40, not greater than 0.45. No family study could be carried out for this variant. The variant allele was tentatively called *BF*SG1* (S Guangzhou 1).

The another variant type SFG2 was encountered twice in the unrelated subjects. The variant band had mobility slightly slower than that of BF F in our electrophoretic system (Fig. 1, lane 4). It is similar to, but considered to be slightly different from, BF F03 described by Dykes *et al.* (1983). It was possible for one of the propositi to carry out family study. As shown in Fig. 2, four members were found to carry the variant allele tentatively called *BF*FG2* (F Guangzhou 2), indicating autosomal, codominant inheritance.

To our knowledge, at least 16 BF variants have been described besides the polymorphic allotypes, S, F, F1 and S07. Majority of these rare variants have been found in Caucasoid or Negroid populations. As for Mongoloid populations, a single rare variant, BF FT(F075) has been reported in Japanese (Tokunaga *et al.*, 1981, 1982). Recently, another variant, BF S065, was reported from Japanese, but the details of this variant were not known (Miyano *et al.*, 1986). In the present study, two further rare variants are added to the list of known BF variants in Mongoloid populations.

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