

Kasuistiken / Casuistics

A Very Slow BF Variant (S085) Detected in a Japanese Population

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Summary. Using high-voltage agarose gel electrophoresis and immunofixation a very slow BF variant was detected in a Japanese person living in Yamanashi district. The family analysis suggested the hereditary occurrence of a new allele, BF*S085.

Key words: Polymorphism, serum groups – Serum groups, BF– BF*S085, new allele

Zusammenfassung. Mittels Hochspannungs-Agarosegelelektrophorese und Immunfixation wurde eine sehr langsam wandernde BF-Variante bei einer im Bezirk Yamanashi lebenden japanischen Person gefunden. Die Familienanalyse zeigte das Vorkommen eines neuen Allels, BF*S085, und dessen Erbllichkeit.

Schlüsselwörter: Polymorphismus, Serumgruppen – Serumgruppen, BF – BF*S085, neues Allel

Introduction

The genetic polymorphism of human properdin factor B (BF) was first described by Alper et al. (1972). It consists of two common alleles, BF*S and BF*F, and two less common alleles, BF*S1 and BF*F1. BF*S1 was later on called BF*S07 according to the nomenclature proposed by Mauff et al. (1978) based on the relative electrophoretic mobility.

Besides these alleles, a number of rare variants have been reported so far: F155 by Mauff et al. (1975); F055 by Hauptmann et al. (1976); F065, F085, and S03 by Mauff et al. (1976); S045 by Hauptmann et al. (1977); S025 by Scherz et al. (1978); F075 by Dykes et al. (1981); F129 by Larsen et al. (1981); F08 by

Davrinche et al. (1982); F135 by Nerl and O'Neill (1982); F02, F03, F07, F11, and F13 by Dykes et al. (1983); F025 by Suzuki and Matsumoto (1986); SG1 and FG2 by Luo et al. (1987).

This paper reports a new BF variant detected in a Japanese individual using high-voltage agarose gel electrophoresis followed by immunofixation.

Materials and Methods

Serum samples were collected from 527 unrelated Japanese individuals living in a central part of Japan, Yamanashi Prefecture, and stored at -20°C until use.

High-voltage agarose gel electrophoresis was carried out as described by Teisberg (1970). Immunofixation was performed according to Alper and Johnson (1969) using specific anti-BF serum (Atlantic Antibodies, USA). The antiserum was diluted 1:1 with physiologic saline and layered over the gel surface. The gel was incubated in a moist chamber for 1 h at 37°C and then washed in physiologic saline overnight.

Results and Discussion

A new BF variant was detected in a 22-year-old woman among 527 Japanese individuals. As shown in Fig. 1, the variant exhibited two major components: the S band and a new band which migrated far more slowly than any other variant

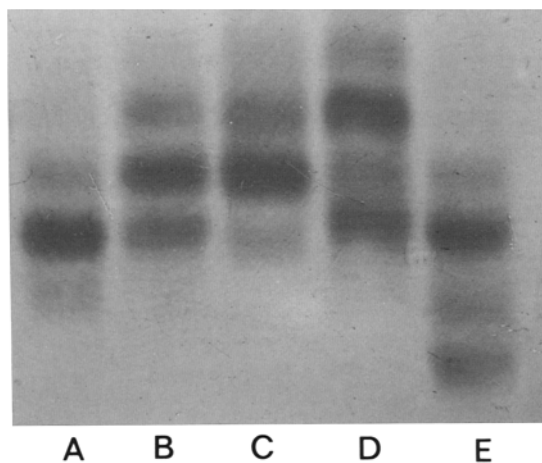


Fig. 1. Agarose gel electrophoretic patterns of BF types. A: BFS; B: BFFS; C: BFF; D: BFF075S; E: BFSS085. The anode is at the top

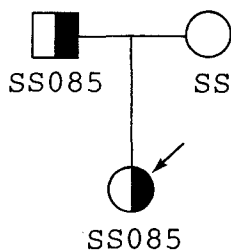


Fig. 2. Pedigree of the family with BF*S085 allele. Arrow indicates the proband

Table 1. Distribution of BF types in a Japanese population

Phenotype	No. observed (%)		No. expected
S	334	(63.4)	336.4
FS	172	(32.6)	167.6
F	19	(3.6)	20.9
F075S	1	} (0.4)	1.7
SS085	1		
Others	0	(0.0)	0.4
Total	527	(100.0)	527.0

Allele frequencies: BF*S = 0.799; BF*F = 0.199; BF*Rare (the combined frequency of BF*F075 and BF*S085) = 0.002

$\chi^2 = 0.759$, $df = 3$, $0.9 > P > 0.8$

so far reported. This new band was named S085 by comparison to the migration difference between the S and F075 bands instead of the difference between the S and F1 bands according to Suzuki and Matsumoto (1986). The reference serum sample of F075S type was kindly provided by Dr. H. Nishimukai, Ehime University School of Medicine. The family study confirmed that the BF*S085 allele was genetically transmitted from her father (Fig. 2).

The results for the distribution of BF types in our population sample are given in Table 1. The observed numbers are in good agreement with the numbers expected according to the Hardy-Weinberg equilibrium. It is of anthropologic significance that in white people BF*F1 and BF*S07 occur with appreciable frequencies (Alper et al. 1972; Mauff et al. 1975; Dykes et al. 1981), whereas these alleles seem absent in Japanese.

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