163. A Japanese Example of the Bombay Phenotype

By Hideo YAMAGUCHI,^{*)} Yasuto OKUBO,^{**)} Tadao TOMITA,^{**)}
Taiko SENO,^{**)} and Masayoshi TANAKA^{**)}

(Comm. by Ren KIMURA, M. J. A., Dec. 12, 1975)

The propositus, Mr. On., was a healthy 48-year-old Japanese whose blood was found to show nonconformity with the Landsteiner's rule when tested at the Gifu Health Care Center. His blood specimen was soon sent to us for further examination.

His red cells gave agglutination reaction with neither of the various anti-A, anti-B, anti-A+B nor anti-H tested. The anti-H reagents employed here were as follows:— human sera from one O_h and one A_h, one eel serum and two lectins, *Ulex europaeus*, *Laburnum alpinum*.

Repeated attempts to make eluates from the propositus' cells of any specificity using anti-A, anti-B, anti-A+B and anti-H (*Ulex* lectin), were unsuccessful, though the genotype of the propositus became evident by the pedigree studies, as will be mentioned below.

Four samples of anti-I from I-negative adults agglutinated Mr. On.'s red cells as well as the control O, I-positive cells. Mr. On.'s cells showed no unusual blood group activities other than ABH (Table I).

As was expected, the propositus' serum contained anti-H of

Table I. Blood Groups of On. Family

	ABO	MNSs	P	Rh	Kk	Le	Fy	Jk	I
II-2	O	MNss	1	ccDEE	kk	a-b-	a+b-	a+b+	+
II-3	O _h	MNss	1	CcDEe	kk	a+b-	a+b-	a-b+	+
II-5*	O _h	MNss	1	CCDee	kk	a+b-	a+b-	a+b+	+
II-6	O	MNss	2	CcDEe	kk	a-b+	a+b-	a+b-	+
II-7	A ₁	MNss	1	CcDEe	kk	a+b-	a+b-	a+b+	+
II-8	A _x B	MNss	1	CCDee	kk	a-b+	a+b-	a+b-	+
II-9	A ₁	MNss	1	CcDEe	kk	a-b-	a+b-	a+b-	+
III-1	A ₁	NNss	1	ccDEe	kk	a-b+	a+b-	a-b+	.
III-2	O	MNss	2	CcDEe	kk	a-b+	a+b-	a-b+	.
III-3	A ₁	MNss	1	CCDee	kk	a-b+	a+b-	a+b+	+
III-4	O	MNss	2	CCDee	kk	a-b+	a+b-	a+b-	+
III-5	A ₁	MNss	1	CcDEe	kk	a-b+	a+b-	a+b+	.
III-6	A ₁	MNss	2	CcDEe	kk	a-b+	a+b-	a+b-	.

*) Propositus

*) Osaka Red Cross Hospital and Osaka Red Cross Blood Center, Osaka.

**) Osaka Red Cross Blood Center.

rather high titer (1:256) which was completely inhibited by O secretor saliva. Absorption of the serum by O, Le (a+) cells left a trace of anti-Le^b besides anti-A of 1:16 and anti-B of 1:128. The titers shown here were those given by anti-globulin test, while in saline they were lower by one or two tubes. These antibodies other than the anti-Le^b were as active at 37°C as at room temperature.

The propositus' saliva did not contain any detectable amount of A-, B-, H- or Le^b-substance, but Le^a-substance of normal level was present in the saliva.

These serological findings clearly demonstrate that the propositus has the Bombay (O_h) condition.

Pedigree studies revealed that one of the four sibs of the propositus (II-5, Fig. 1) was O_h. As the propositus with an O group spouse (II-6) produced two children of A₁ and O group (III-3, III-4), the genotype of the propositus was shown to be A₁O. We could not guess the genotype of the Bombay brother (II-3) because of his deceased spouse. It was noteworthy that all the sibs of the propositus were non-secretor, and that a carrier of variant A_xB (II-8), quite different from O_h happened to co-occur in this pedigree, though both of the variants were extremely rare in Japanese population. The parents of the propositus were related, more detailed information being unavailable.

Fig. 1

Remarks. Taking account of the work by Lanset *et al.* (1) who serologically identified the suppressed antigen of the O_h cells by fixation-elution tests, our attempts to make eluates from the cells of both the propositus and his O_h brother were repeatedly done but with negative results. Fortunately, the pedigree studies made this point clear. The carriers of the Bombay phenotype in the present pedigree

were usual in that their red cells gave the reaction Le (a+b-) (Table I).

In Japan the Bombay phenotype must be extremely rare, because since in 1970 the first example of this phenotype was reported by Iseki *et al.*,²⁾ there has been no published case until the appearance of the present example. The case described by Iseki *et al.*²⁾ seemed quite typical in serological findings, but the suppressed antigen of their case remained uncertain.

On the other hand, several reports on the Bombay-like or para-Bombay phenotypes have been read at the recent annual meetings of the Japan Society of Blood Transfusion, and almost all of them seemed to belong to "row III" classified by Race and Sanger (1975).³⁾ We^{4),5)} ourselves have published two such para-Bombay cases. For the background of these para-Bombay phenotypes we⁵⁾ suggested a recessive regulator gene *es* (generally called *z*) which is opposite in its effects to the non-secretor gene, *se*. Recently Kojima *et al.*⁶⁾ and we⁷⁾ encountered respective examples of A_h which fell under "row II" by Race and Sanger.³⁾

At any rate, sometimes to us occurs the idea that some people homozygous for both a rare allele at the *Zz* locus and the non-secretor gene *se* might not be phenotypically distinguished from people homozygous for a third allele at the *Hh* locus, or even from the O_h people.

Acknowledgement. We wish to express our thanks to Prof. Ren Kimura for his revision of this manuscript, to Prof. Takehiko Kikuchi for his constant encouragement, and to Miss Yasuko Shibuya (Gifu Health Care Center) for collecting blood specimens.

References

- 1) Lanset, Suzaenne, Ropartz, C., Rousseau, P.-Y., Guerbet, Y., and Salmon, C.: Une famille comportant les phénotypes Bombay: O_h^{AB} et O_h^B. *Transfusion*, Paris, **9**, 255 (1966) cited by Race, R. R., and Sanger, Ruth: *Blood Groups in Man*, 6th ed. (1975).
- 2) Iseki, S., Takizawa, H., and Takizawa, Haruko: Immunological properties of "Bombay" phenotype. *Proc. Japan Acad.*, **46**, 803 (1970).
- 3) Race, R. R., and Sanger, Ruth: *Blood Groups in Man*. 6th ed. Blackwell Scientific Publications (1975).
- 4) Kitahama, M., Yamaguchi, H., Okubo, Y., and Hazama, F.: An apparently new B_h-like human blood type. *Vox Sang.*, **12**, 354 (1967).
- 5) Yamaguchi, H., Okubo, Y., and Tanaka, M.: Co-occurrence of A_m^h and B_m^h blood in a Japanese family. *Proc. Japan Acad.*, **48**, 629 (1972).
- 6) Kojima, K., Otte, T., and Matsukura, H.: One example A_h. *J. Jap. Soc. Blood Transf.*, **20**, 28 (1974) (in Japanese).
- 7) Okubo, Y., Yamaguchi, H., Tomita, T., and Tanaka, M.: A_h phenotype in a family. *Japan. Red Cross Med. J.*, **27**, 49 (1975) (Abstract in Japanese).