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Para-Bombay phenotype: A case report from a tertiary care hospital from South Gujarat

Jitendra N. Patel, Ajaykumar B. Donta¹, Arpit C. Patel, Amrish N. Pandya, Swati S. Kulkarni¹

Abstract:

The blood specimen of a 30-year-old male donor showing a discrepancy in cell and serum grouping was targeted for detailed study at the blood bank at tertiary care hospital in South Gujarat. Forward grouping showed group as "O" RhD positive and reverse grouping as group "A." Further testing confirmed that the individual's blood group was para-Bombay A (para-AH). Family members were screened, and younger brother was also identified as para-Bombay phenotype. The para-Bombay phenotype is very rare, and only a few cases have been reported from India. This blood group is characterized by the absence of ABH antigens on red blood cells (RBC's) with the presence of ABH substances in body secretions or by the weak expression of ABH antigens on RBC's with the absence or presence of substances in body secretions. This rare phenotype can be mislabeled as "O" if all detailed investigations are not performed.

Keywords:

H-antigen, para-Bombay, secretor status

Introduction

The classic Bombay phenotype is rare blood type which was first reported in 1952 in Bombay, India and is associated with the ABO and H blood group systems.^[1] In Bombay phenotype, both red cells and secretions are deficient in H, A, and B antigens. In routine blood testing, cell grouping shows characteristics of O group. Serum grouping reveals the presence of anti-H activity in the serum in addition to the natural ABO antibodies. The anti-H present is of clinical significance since it can activate complement and cause hemolysis. As their blood is compatible only with the Bombay phenotype cells, getting blood unit for transfusion is not easy. Bombay phenotypes have a relatively higher incidence in India.^[2] Its prevalence

is reported as 1: 4600 in Ratnagiri district,^[3] 1: 7600 in Mumbai,^[4] 1 in 10,000 in India, and 1/1,000,000 individuals in Europe.^[5,6]

Another related phenotype is para-Bombay which is extremely rare, with only a very few cases being reported and none of the published studies have reported the prevalence of para-Bombay phenotype in India. However, in the Chinese population, more number of para-Bombay than Bombay phenotype cases have been reported.^[7] The para-Bombay phenotype is characterized by the absence of ABH antigens on red blood cells (RBCs) with the presence of ABH substances in body secretions or by the weak expression of ABH antigens on RBCs with the absence or presence of substances in body secretions. In contrast, Bombay phenotypes have an absence of ABH antigens on both, the RBCs as well as in body secretions.^[8] Furthermore in the para-Bombay phenotype, the atypical

Department of Immuno-Hematology and Blood Transfusion, Government Medical College and New Civil Hospital, Surat, Gujarat, ¹Department of Transfusion Medicine, ICMR- National Institute of Immunohaematology, Mumbai, Maharashtra, India

Address for correspondence:

Dr. Swati S. Kulkarni, National Institute of Immunohaematology (ICMR), 13th Floor, NMS Building, KEM Hospital Campus, Parel, Mumbai - 400 012, Maharashtra, India. E-mail: swatiskulkarni@gmail.com

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antibodies in the serum react preferentially at lower temperatures (below 20°C–22°C).^[9]

Case Report

A 30-year-old healthy individual, after obtaining informed consent had donated blood in voluntary blood donation camp and ABO grouping was performed using standard serology techniques.^[10] Testing of donor's red cells with commercial monoclonal anti-A and anti-B reagents (Tulip Diagnostics Pvt Ltd., and Span Diagnostics, India) showed no detectable A and B antigens (O group) by tube method. The red cells also showed the presence of RhD antigen with two different commercial anti-D reagents. Reverse/serum grouping with in-house A, B, and O pooled cells showed the presence of anti-B antibodies in serum (A group). Cell and serum grouping results were concordant by tube and column agglutination technique (Tulip Diagnostics Pvt Ltd., India). To resolve the discrepancy between cell and serum grouping, further immunohematology workup was carried out. Testing the sample with commercially available anti-H-lectin showed the absence of H antigen on red cells, but no anti-H activity was detected by reverse grouping and by titration for the naturally occurring anti-H at different temperatures (37°C, 22°C and 4°C). Adsorption-elution test showed the absence of A, B, and H antigens. Test for secretor status showed the presence of A and H substances in the saliva of donor, thus diagnosing the present case as para-Bombay A phenotype (para-A_H). The donor's serum agglutinated Group B red cells showing a titer of up to 1:8 at 22°C, 1:4 at 37°C and 1:16 at 4°C but showed no reaction with A and O red cells. The serological workup is summarized in Table 1. Donor's family members were also tested, father was O RhD positive, mother A RhD positive and his younger brother (27 years) showed same reactivity pattern as propositus in all the serological tests performed and was hence labeled as para-Bombay phenotype (Para-A_H).

Discussion

As H antigen is the precursor for the A and B antigens, it is expressed on all red cells except in the rare Bombay and para-Bombay phenotype showing its absence or deficiency. The ABO genes determine the presence of A and B antigens, whereas the H antigen is a result of α -(1,2)-fucosyltransferase (*FUT*) genes. *FUT1* (*H* gene) determines the presence of H antigen on the RBCs and *FUT2* (*Se* gene) in body secretions. *FUT1* forms the H antigen which is preferentially expressed in erythroid tissues and vascular endothelial cells by fucosylation of the type 2 chain oligosaccharides on red cell glycoproteins and glycolipids. *FUT2* recognizes type 1 chain precursors to form H type I antigen in secretions and tissues such

Table 1: Pattern of reactions observed with cells, serum and saliva

	Reaction grade*	Interpretation
Cell grouping		
Anti-A	0	O Rh (D) positive
Anti-B	0	
Anti-AB	0	
Anti-A1	0	
Anti-D	4+	
Anti-H	0	H antigen negative
Serum grouping		
A cells	0	A group
B cells	4+	
O cells	0	
O cells + Enzyme	0	
Donor's Anti-B titre at different temperature (°C)		
37	1:4	Higher titre at
22	1:8	lower temperature
4	1:16	
Adsorption elution test		
Anti-A	0	Absence of A, B
Anti-B	0	and H antigens on
Anti-AB	0	RBCs
Secretor status		
A cells	0	H and A substance
B cells	3+	present in saliva
O cells	0	

*0 = No agglutination, 1+ = Multiple small agglutinates with hazy supernatant, 2+ = Multiple large agglutinates with clear supernatant, 3+ = 2–3 large agglutinates with clear supernatant, 4+ = Single large agglutinate. RBC = Red blood cells

as secretory glands and digestive mucosa. Bombay phenotype is characterized by the absence of ABH blood group antigens both on the surface of RBCs and in saliva resulting from both silenced mutations in *FUT1* (*h/h*) and *FUT2* (*se/se*) genes.^[7] All the Bombay phenotype cases reported in India showed only the T725G mutation of *FUT1* and the 10 kb gene deletion of *FUT2* which are responsible for this rare phenotype, suggesting its unicentric origin.^[11]

The para-Bombay phenotype results from a silenced *FUT1* gene (*h/h*) but an active *FUT2* (*Se/Se* or *Se/se*) gene to synthesize H Type I antigen (and A/B antigens) in the secretions (H-deficient secretors) that may be adsorbed onto RBCs from the plasma or from a mutated *FUT1* gene resulting in great diminished enzyme activity to produce low amounts of H Type II antigen (and A/B antigens) on the surface of RBCs, which could only be detected by adsorption and elution technique.^[7]

Unlike Bombay, the para-Bombay blood group individuals can express type 1 chain A, B, H antigens in their plasma and secretions. The weak A and/or B antigen expression is due to passively adsorbed antigens on red cells.^[8] The donor test results with red cells, serum,

and saliva revealed blood group was para-Bombay A_H as described in Table 1. A similar case of a 54-year-old male anemic patient with the discrepancy in cell and serum grouping was reported as para-Bombay B (Para B_H) from Andhra Pradesh after detailed immunohematology workup.^[12]

Para-Bombay blood group individuals usually retain some H antigen on RBCs and weak anti-H activity, which is often demonstrable only at 4°C or by using adsorption and elution techniques.^[8,9] A case of partial suppression where antigens were detected by elution tests and unlike classical Bombay type, normal amount of appropriate blood group substances was present in saliva have been reported earlier.^[13] In the present study, A or B antigens expression in donor was not detected by adsorption–elution and anti-H activity was not revealed either by routine techniques or at 4°C. The donor being para-Bombay phenotype was serologically revealed only after testing red cells with anti-H lectin and special tests like ABH titer, secretor status along with cell and serum grouping.

Conclusion

This case of para-Bombay phenotype was detected as a result of discrepancy in cell and serum grouping. This case highlights the importance of proper blood grouping (cell and serum both) and use of anti-H reagent for the identification of para-Bombay phenotype, otherwise this donor would have been mislabeled as Group O. In routine blood banking, besides blood grouping detailed serological tests such as secretor status, adsorption-elution, and serum antibody titration at different temperatures should be performed. The authors also recommend molecular testing for further confirmation in such cases.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will

be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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