Case Report:

Para-Bombay phenotype: report of a rare blood group

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ABSTRACT

The blood sample of a 54-year-old male patient who presented with signs and symptoms suggestive of anaemia was submitted to the Blood Bank for blood grouping and cross-matching. In forward grouping, no agglutination was observed with A, B and AB antisera, but agglutination was noticed with D antiserum (Group O). In reverse grouping, there was agglutination in tube labelled A and no agglutination in tubes B and O (Group B) resulting in discrepancy between forward and reverse grouping. Further testing confirmed that the individual's blood group was Para-Bombay B (Para-BH), which is a rare entity. The Para-Bombay phenotype is very rare. Only a few cases of Para-Bombay were reported in India till now and none from Andhra Pradesh. This entity is characterized by the absence of H, A and B antigens on the red cells but their presence in saliva and secretions of gastrointestinal and genitourinary tracts. Proper identification of this phenotype is very important; otherwise this particular blood group may be mislabelled as group O.

Key Words: Para-Bombay, H-antigen, Secretor status

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INTRODUCTION

The H antigen is the precursor for the formation of A and B antigens and its absence is termed as H antigen deficient phenotype.[1] It results in, Bombay or Para-Bombay blood group in an individual. The H blood-group-deficient phenotypes have been found in diverse ethnic groups/nationalities, with a much higher frequency being noted in Lahu Chinese (2.2%) than reported in any other ethnic group.[1] In India its prevalence has been estimated to be 1 in 10,000.[2]

The Para-Bombay phenotype is very rare, with only a few cases being reported in India[3] till now; to the best of our knowledge no case has been reported from Andhra Pradesh. Para-Bombay phenotype is characterized by the deficiency of H, A and B antigens on the red cells. These persons inherit hh/Sese or hh/SeSe genes. Though they lack H antigen on RBCs, it is present in secretions and hence these patients are referred to as "Para-Bombay secretors" or "red blood cell (RBC) H negative secretors",[3] in distinction to "Bombay phenotype" which refers to individuals whose RBCs and secretions lack the H antigen.[4]

CASE REPORT

A 54-year-old male patient attended to our hospital with signs and symptoms suggestive of anaemia. The investigations revealed a haemoglobin of 4.9 g/dL; RBC count 1.46 millions/ mm^3; packed cell volume 17 per cent; total leukocyte count 2,700/ mm^3; differential leukocyte count neutrophils 44%, lymphocytes 45%, eosinophils 7%, and monocytes 4%; erythrocyte sedimentation rate 110 mm at the end of first hour; mean corpuscular volume 99 fL; mean corpuscular haemoglobin 28 pg and platelet count 126,000/mm^3. Peripheral smear examination showed mild to moderate anisopoikilocytosis with predominant normocytic normochromic morphology admixed with a few macrocytes, occasional macro-ovalocytes and polychromatophils; haemparasites were absent, leukopenia with relative lymphocytosis, eosinophilia and thrombocytopenia. Plasma glucose, serum lactate dehydrogenase (LDH), ferritin and creatinine

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were within normal limits. A blood sample obtained from the patient was submitted to the Department of Immuno-Haematology and Blood Transfusion for blood-grouping and cross-matching, with a request for issue of 3 units of packed red blood cells. ABO grouping was performed using standardized serological techniques.[5]

Testing of patient's red cells showed no detectable ABO antigens on forward/cell grouping (O group). Reverse/serum grouping showed presence of A antibodies in serum (B group) (Table 1).To resolve discrepancy between cell and serum grouping, Immuno-haematology work up was carried out. Testing the sample with an in-house Anti-H serum and commercially available H-lectin (prepared from plant *Ulex europeus* extract) showed no agglutination reaction with red cells. Secretor status was also done to assess the presence of soluble blood group substances, which showed presence of B and H antigens in saliva, thereby the present case was diagnosed as Para-Bombay B phenotype (Para-B_H). Further work-up was not possible as the patient was discharged without receiving any blood transfusion and he was lost to follow-up.

DISCUSSION

The H antigen is ubiquitously expressed on all red cells except in case of the rare Bombay phenotype. The H antigen is the precursor of the A and B antigens on red blood cells. The ABO locus determines the A and B antigens, whereas α-(1,2)-fucosyl transferase (FUT) genes, FUT1 (H gene) and FUT2 (Se gene) determine the H antigen, the precursor of A and B antigens.[6] The two different α-(1-2)-fucosyl transferase enzymes encoded by two closely linked genes on chromosome 19q13.3. FUT1 specifically fucosylates type 2 chain oligosaccharides on red cell glycoproteins and glycolipids to form H antigen. In contrast FUT2 recognizes type 1 chain precursors to form type 1H antigen in secretions. The FUT2 is not expressed in red cells but is expressed in salivary glands, gastrointestinal and genitourinary tissues. The Para-BH phenotype individuals are H-deficient secretors. Genetically, these individuals are homozygous for a non- functional H gene (hh), but they inherit at least one functional secretor genes (Se). The red cells from H-deficient secretors lack serologically detectable H-antigen but can carry small amounts of A and/or B antigen

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

	Reagent	Reaction grading	Interpretation
Cell grouping	Anti-A	0	
	Anti-B	0	
	Anti-AB	0	O Rh(D)positive
	Anti-D	4+	
Serum grouping	A_1 cells	4+	
	B cells	0	B group
	O cells	0	
Test for H antigen	Anti-H (in-house)	0	
	Anti-H lectin (commercial)	0	H antigen negative on red cells
Saliva secretor status	A_1 cells	2+	
	B cells	0	Type 1H, B antigen
	O cells	0	present in saliva

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

because unlike classic Bombay blood group, Para-BH blood group persons express type 1 chain A, B, H antigens in their secretions and plasma. These antigens are passively adsorbed onto red cells, resulting in weak A or B antigen expression. Para-Bombay blood group individuals usually retain some H antigen on RBCs and weak anti-H activity, which is often demonstrable only at 4 ^{o}C or by using adsorption and elution techniques. In our patient, no anti-H activity was demonstrated either by routine techniques or at 4 ^{o}C. Absence of H, A, and B antigens on red cells and secretions is Bombay phenotype. Genetically Oh individuals are homozygous for nonfunctional H (hh) and Secretor (sese) genes.[5]

Problems may arise in finding compatible units for these patients because of anti-H or anti-IH, but most often these are not clinically significant. Therefore, when whole blood units of normal ABO blood groups compatible by indirect anti-globulin test (IAT) are transfused, the survival is expected to be almost normal. These weak isoagglutinins may not be very clinically significant and it was suggested that when Para-Bombay blood is not available, the compatibility testing for Para-Bombay A persons should be performed with group A and group O packed red blood cells (RBC); Para-Bombay B with group B and O packed RBC; Para-Bombay AB with group A, B, AB and O packed RBC. For cross matching, the indirect anti-globulin test by pre-warmed technique should be used.[7]

Without the use of anti-H lectin or serum, this particular patient might have been labelled as group O rather than Para-BH phenotype. This case stresses the importance of judiciously using anti-H in blood grouping.

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