ISSN : 2309-3234

TMSS Medical College Journal (TMCJ)

Volume 20, No. 01, January 2023

Contents

An Official Publication of TMSS Medical College

TMSS Medical College Journal (TMCJ)

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An Official Publication
of
TMSS Medical College, Bogura, Bangladesh

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The TMSS Medical College Journal (TMCJ) is published two times a year (January and July). The TMSS Medical College Journal (TMCJ) is approved by Bangladesh Medical & Dental Council (BM&DC/4-D-2016/1478: Dated 15.01.2017). It accepts original articles, review articles and case reports. While every effort is always made by the Editorial Board and the members of the Journal Committee to avoid inaccurate or misleading information appearing in the TMSS Medical College Journal, information within the individual article are the responsibility of its author(s). The TMSS Medical College Journal, its Editorial Board and Journal Committee accept no liability whatsoever for the consequences of any such inaccurate and misleading information, opinion or statement.

Printed by

TMSS Printing Press
Bogura, Bangladesh
e-mail: tmssprintingpress@gmail.com

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Case Report

Bombay Phenotype- A Rare Blood Group Detected First Time in TMSS Medical College Hospital, Bogura: A Case Report

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Abstract

Bombay phenotype (Symbol Oh), popularly well-known as Bombay blood group was first discovered by Y M Bhende et al in 1952 at Bombay (now known as Mumbai). It is a rare phenotype which shows absence of A, B, H antigens on red cell membrane and presence of anti-A, anti-B and anti-H antibodies in serum. H antigen on RBCs is the precursor structure on which *ABO* genes act to produce A and B antigens. As H antigen is absent, no A and B antigens are produced. Bombay phenotype (Oh) lacks normal expression of the A, B and H antigens because of the inheritance of the hh genotype. Their blood is incompatible with any other blood groups, except blood of another Bombay if they need blood transfusion. Prevalence of Oh is 1 per 10,000 in India and 1 per 1,000,000 in Europe. In Bangladesh, it is 1 per 0.006%. We found a first case of Bombay phenotype at TMSS Medical College, Bogura, which was confirmed by testing of blood and saliva using anti-H lectin.

Key words: A Rare Blood Group, Antigen, Bombay phenotype.

Introduction

Bombay phenotype (Symbol Oh), popularly well-known as Bombay blood group was first discovered by Y M Bhende et al in 1952 at Bombay (now known as Mumbai), India. This is the reason why this type of blood got the name Bombay phenotype. It is a rare phenotype which shows absence of A, B, H antigens on red cell membrane and presence of anti-A, anti-B and anti-H antibodies in serum.[1] The molecular basis of this blood group has been proven to be mutations of the FUT1 gene (H gene) which results in the formation of complete H-deficient phenotype (genotype hh).[1, 2]

After the cloning and characterization of the FUT1 gene in 1990 and proving that the gene was the molecular basis of the Bombay phenotype in 1994, the H blood group system was established. The H blood group system, ISBT symbol H (018), consists of a single antigen (H) defined by a terminal fucose residue found on red blood cells and in secretions.[3]

The H gene must be inherited to form ABO antigens on the RBCs. Inheritance of the H gene (genotype HH or Hh) results in the formation of the H antigen. The H antigen on RBCs is the precursor structure on which ABO genes act to produce A and B antigens. If H antigen is absent, no A and B antigens are produced. The term Bombay has been used to refer to the phenotype that lacks normal expression of the A, B and H antigens because of the inheritance of the hh genotype.[4]

Bombay phenotypes are commonly mistaken as O group. As like O group, it has no A and B antigen but differs from O group by lacking H antigen on RBC. In RBC testing using anti-A and anti-B, the Bombay would phenotype as O blood group but do not react with the anti-H lectin (Ulex europaeus), whereas normal O group individual react strongly with anti-H lectin. The Bombay anti-H can often be potent and reacts strongly at 37°C. It is an IgM antibody that can bind complement and cause RBC lysis. Transfusing normal group O blood (with the highest concentration of H antigen) to a Bombay recipient (anti-H in the serum) would cause immediate severe hemolytic transfusion reaction, which can be fatal and even lead

to death. Therefore, only blood from another Bombay individual will be compatible and can be transfused to a Bombay recipient.[4, 5]

This People who lack the H antigen do not suffer from deleterious effects and H-deficient is only an issue if they need a blood transfusion, because they would need blood without the H antigen present on red blood cells i.e. from another Bombay phenotype.[5] Bombay individuals can donate red cell concentrate (RCC) to any ABO group if Rh is compatible. They can receive fresh frozen plasma (FFP) and cryoprecipitate from any group but can receive red cells only from other Bombay individuals.[6]

About 99.9% of all individuals have an HH or Hh genotype in the world.[2, 4, 6] So the incidence of Bombay phenotype is 0.1%. This very rare phenotype is generally present in about 0.0004% (about 4 per million) of the human population, though in some places such as Mumbai locals can have occurrences in as much as 0.01% (1 in 10,000) of inhabitants.[5, 6] In Bangladesh, the first documented Bombay phenotype was found in the Miah family in 1990 at Narayanganj.[7] Now sporadic cases are identified in different laboratories where the facilities are available to confirm Bombay phenotype.

Case Report

A healthy male of 30 years came to our Transfusion Medicine Department for blood grouping as a routine health checkup including other laboratory tests as a prerequisite of joining a job. Although he told that his blood group is O positive and he justified his saying showing a blood report done from another tertiary care hospital.

Firstly, we noticed the anomaly in routine testing using forward grouping (cell grouping) and reverse grouping (serum grouping) and suspect the case as Bombay (Oh) phenotype. We confirmed this sample as Bombay phenotype by testing with anti-H lectin. Finally, we tested patient's saliva (hemagglutination inhibition test) and found that he is a non-secretor i.e., his body secretion does not contain ABO antigens which also confirmed the diagnosis of Bombay phenotype. Other pathological reports were all within normal limit.

Discussion

In our country, most of the private blood centers perform blood grouping using one slide method (Only cell grouping), but where transfusion specialists are present, whether in private or govt. blood transfusion centers, mostly double slide methods i.e., both cell grouping and serum grouping are used, although tube method is the best. If one slide method is used, incorrect grouping may occur, thus missing Bombay blood group. For confirmation of ABO grouping, we should do both cell grouping using anti-A, anti-B and anti AB against patient's cells and also serum grouping using A cell, B cell and O cells against patient's serum. During blood grouping, positive reaction with O cell in reverse grouping gives a clue to Bombay phenotype. Normally O cell has no A or B antigens but has potent H antigens which will react with anti-H of Bombay blood group.

Literature review revealed Bombay phenotype is more prevalent in closed communities with a higher rate of consanguineous marriage. It is very likely that other family members could carry the same phenotype. A study found the frequencies of Bombay phenotype 1 per 8,000 in Taiwan, 1 per 10,000 in India, and 1 per million in Europe.[2] A study in India revealed that out of 179 cases, incidence of Bombay phenotype in Maharashtra is the highest i.e., 62.6% followed by 7.8% in Karnataka, 4.5% in Andhraprodesh, 4.5% in Goa and then followed in other states.[6] Another report in India showed that in the states of Andhra Pradesh, Tamil Nadu, and Karnataka, prevalence of the Bombay phenotype is 0.048%, 0.004%, and 0.005%, respectively.[8]

No specific statistics of people with Bombay blood group in Bangladesh is available. A recent study in Bangladesh revealed that prevalence of Bombay Phenotype is 0.006%.[9]

In our case, father of the man belongs to blood group B group and mother belongs to O group. His blood group and also his only sister's blood group are supposed to be B or O but he is a Bombay and his sister is group O. We assumed that both of his father and mother are heterozygous for H gene i.e., their

genotypes Hh and they inherit hh to their son as he is Bombay and Hh or HH to their daughter as she is of group O. There was no consanguine history of marriage of his family. No other near relative was found to be of Bombay phenotype.

General Characteristics of Bombay Blood Group: a) Absence of H, A, and B antigens; no agglutination with anti-A, anti-B, or anti-H lectin, b) Presence of anti-A, anti-B, anti-A,B, and a potent wide thermal range of anti-H in the serum, c) A, B, H non-secretor (no A, B, or H substances present in saliva), d) Absence of α-2-L-fucosyltransferase (H enzyme) in serum and H antigen on red blood cells, e) Presence of A or B enzymes in serum (depending on ABO genotype). f) A recessive mode of inheritance (identical phenotypes in children but not in parents). f) RBCs of the Bombay phenotype (Oh) will not react with the anti-H lectin (Ulex europaeus), g) RBCs of the Bombay phenotype (Oh) are compatible only with the serum from another Bombay individual.[2, 4]

Conclusion

Compulsory use of cell grouping and serum grouping and in suspected cases of Bombay phenotype, use of anti-H lectin and detection of secretor status; all are important tools for detection of Bombay phenotype. As this condition is very rare, any person of this phenotype who needs an urgent blood transfusion will probably be unable to get Bombay blood, as no blood bank would have it in stock. So every blood bank should have registry of individuals of Bombay group which may help to find these individuals to get blood in this situation.

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(TMSS Medical College Journal 2023; 20(1): 41-43)