

Popular Article

Bombay (Hh) Blood Group: A Rarest Type of Blood Group System

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Introduction

'Bombay blood group' the rarest blood group, which was initially identified by Dr. Y.M. Bhide in 1952 in Bombay (now known as Mumbai) is an extremely uncommon blood type that typically makes up 0.0004% (or 4 million) of the world's population. However, in Mumbai, the blood type can make up as much as 0.01% of the population, or 1 in 10,000 people. Normally the HH antigen leads the production of A, B or O antigen. The O blood group also expresses the H antigen, which is not expressed by those with the hh phenotype. As a result, their red blood cells are unable to produce substance A or substance B, often known as A or B antigen, respectively. People with the Bombay blood group are therefore able to donate blood to any individual in the ABO blood group system, but they are unable to receive blood from any individual in the ABO blood group system. Put otherwise, a safe and problem-free blood transplant from the same phenotype is only possible for persons who possess Bombay Blood. It is challenging to transfuse blood of this phenotype since the blood group is uncommon. Those who have inherited two

recessive strains of the H gene belong to the Bombay Blood group. These people don't make the H carbohydrate. Given that both parents have the allele, their children will inherit this blood type. Furthermore, there is a greater likelihood that a child and both parents will have the same blood type with Bombay blood. Noble families are another example of inbreeding based more on tradition than genetic diversity in a given area.

This incident occurred when a patient who needed a blood transfusion was admitted to the KEM hospital. Blood with blood group O was transfused to the patient as a result of the red blood cells in the blood being clustered similarly to that of the O group during the routine blood diagnostic procedure. The blood transfusion was discontinued when the patient eventually experienced a hemolytic transfusion reaction. A new blood type known as the Bombay blood group, also known as the HH group, was later identified after a thorough analysis of the patient's blood was conducted. The H antigen is the main protein found in all blood types. The H antigen serves as the basis for antigens A and B. Therefore, having the A blood group indicates that the individual possesses both type B and type A antibodies in their blood. In a similar

way, an individual with blood type B has type B antigen and type A antibody in their system. While blood group "O" has both type A and type B antibodies but no antigens, blood group AB has both type A and type B antigens in their blood but none of the latter. In India as per 'THE TIMES OF INDIA' report on 11th march 2024, around 450 individual possess the Bombay blood group.

Basic biochemistry behind the Bombay blood group

The biosynthesis of the H antigen and the A and B antigens involves a series of enzymes (glycosyltransferases) that transfer monosaccharides. The end product is an oligosaccharide chain that binds to proteins and lipids that are fixed in the membrane of red blood cells. A particular fucosyltransferase 1 (Gene symbol: FUT1) is responsible for producing the H antigen. The H antigen can be changed into the A, B, or both antigens, depending on the individual's ABO blood type. In individuals with blood group O, the H antigen stays unaltered. Consequently, blood type O has the highest concentrations of the H antigen, whereas blood type AB has the lowest concentrations. The H locus (FUT1) and the Se locus are two genomic areas that encode two enzymes with extremely similar substrate specificities (FUT2). The FUT1 gene, which is expressed in RBCs, is located at the H locus. The production of the H antigen on RBCs requires the presence of at least one functional copy of FUT1 (H/H or H/h). In the event that both FUT1 copies are inactive (h/h), the Bombay phenotype is observed. The Se locus contains the FUT2 gene, which is expressed in secretory glands. Individuals who are "secretors" (Se/Se or Se/se) contain at least

one copy of a functioning enzyme. They produce a soluble form of H antigen that is found in saliva and other bodily fluids. "Non-secretors" (se/se) do not produce soluble H antigen. The enzyme encoded by FUT2 is also involved in the synthesis of antigens of the Lewis blood group.

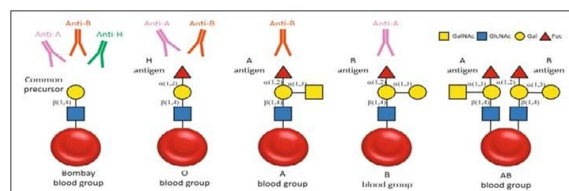


Fig.1 Schematic representation of Bombay, O, A, B, and AB blood groups antigen's carbohydrate epitopes present in RBC surface (RBC are depicted as red circles) and antibodies (Anso et al., 2023).

Molecular analysis

On chromosome 19, the H blood group locus (containing FUT1) and the secretor locus (containing FUT2) are present. FUT1 and FUT2 are closely linked with a little 35 kb distance apart. They most likely originated from a gene duplication of a common ancestor due to their great homology. Four exons covering more than 8 kb of genomic DNA are present in the H locus. Point mutations in the FUT1 gene cause the Bombay phenotypes. The Tyr316Ter mutation in the FUT1 coding area is the source of the conventional Bombay phenotype. The mutation results in a shortened enzyme with a stop codon added, which makes the enzyme inactive because it is missing 50 amino acids from its C-terminal. Numerous mutations may be the cause of the Bombay phenotype in Caucasians.

Conclusion

The Bombay or H blood group system is highly uncommon blood group system in human population. The H antigen is sole responsible for the synthesis of A, B and O antigen and its failure leads to the occurrence of this disease. The person posses this blood group can receive the blood from person having the same blood group. Due to the presence of the only few individuals with this blood group, makes this blood group system highly crucial.