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CASE REPORT

A 50 year old Muslim male patient was admitted to the ICU of Shifa Hospital, Bangalore with marked haematemesis. On examination the patient was jaundiced, moderately febrile, and restless. His BP was low with tachycardia. His ECG was normal. His abdomen showed marked ascites with edema over his feet. US abdomen showed contracted cirrhotic liver with moderate splenomegaly. According to his history, he turned out to be an old case of cirrhosis. His CBC showed marked anaemia (Hb – 4 gms %) with moderate leucopenia and thrombocytopenia. His PT (with INR) was moderately prolonged. His other blood reports showed moderately increased bilirubin, reversal of A/G ratio, moderate increase in AST (SGOT), ALT (SGPT), and Alkaline Phosphatase. There was also a mild increase in urea and creatinine.

(Keywords: Bombay phenotype, transfusion, Oh blood group, rare blood type, blood antigens)

TREATMENT

Immediate treatment was to control his haematemesis (due to oesophageal variceal bleeding) and to improve his anemia. His haematemesis was controlled by giving intravenous injections of Vasopressin (Pitressin) and Somatostatin. Endoscopy was also performed and band ligation of oesophageal varices was performed. His blood was sent to the blood bank for grouping and cross matching. The red cells grouped like the O group, while his serum reacted with all O group cells available in the blood bank during cross matching or compatibility test, making the blood bank official realize that the patient is carrying the rare Bombay Blood Group. Reverse grouping or Serum grouping was performed to confirm this group. The difficulty with the Bombay Blood Group (Bombay phenotype Oh) is that the individuals having this group can either receive autologous donation or blood from an individual of Bombay phenotype only; no other blood will match in case of an emergency blood transfusion. As this blood group is very rare, we could not find a single unit of this blood group in Bangalore and immediate blood transfusion could not be performed. Ultimately, after about 8 hrs, through the help of patient’s relatives we could get with difficulty two units of this blood group from Chennai (Tamil Nadu) and the next day, two units from Hyderabad (Andhra Pradesh). After transfusion the patient’s Hb improved to 8 gms %. The following day we could get two additional units of this blood group from Bombay (Mumbai). After transfusion, the patient’s Hb went up to 10.5 gms%. With the help of diuretics (Lasix + Aldactone) his ascites improved and the patient was discharged after two weeks. At the time of discharge his Hb was 11.5 gms %.

DISCUSSION

The Bombay Blood Group is the rarest of the rare in blood groups. It is so called because Bhende et al. (11) reported it first in Bombay (now called Mumbai), India. People bearing this blood group will not possess A, B, and H antigens in their red cells. They have anti-A, anti-B and anti-H antibodies. Their sera are incompatible with all red cells except those with same rare blood groups. During cell grouping or routine grouping, Bombay Blood Group would be categorized as O group because they wouldn’t show any reaction to anti-A and anti-B antibodies just like a normal O group. When a cross matching with different blood bags of O group is done, then it would show cross-reactivity or incompatibility. Therefore Reverse grouping or Serum grouping has to be performed to detect the Bombay Blood Group. Thus, the patients who test as type O may have the Bombay phenotype if they have inherited two
recessive alleles of the H gene, (their blood group is Oh and their genotype is “hh”), and so do not produce the “H” carbohydrate (fucose) that is the precursor to the “A” and “B” antigens. It then no longer matters whether the A or B enzymes are present or not, as no A or B antigens can be produced since the precursor antigen H is not present.

These individuals were, therefore, genetically termed as homozygous hh or Bombay phenotype. They were non-secretors of ABH and the majority of them were Le (a+) [Balgir (5)]. Watkins and Morgan (31) and Gerard et al. (17), later elucidated the biosynthetic pathway for ABH and Lewis (Le) antigens. Recently, molecular genetic studies were carried out to determine role of the H, Se, and Le genes in the expression of H antigen in secretions and Lewis blood group antigen on erythrocytes [Kaneko et al (19), Oriol et al. (25)]. The H substance (antigen) is a precursor protein from which the blood group proteins are formed.

All human red blood cells, with exceedingly rare exceptions (Bombay Blood Group), carry the red cell H antigen. It is present in greatest amount on type O red cells and least on type A1B cells. This H substance is bio-chemically produced by the binding of Fucose to the surface glycoproteins, the process being catalyzed by Fucosyl transferase. If N-acetyl galactosamine binds to the H substance, it forms the blood group A, whereas if galactose binds to it, it forms the group B. Absence of any binding substance to H produces the O blood group.

The Bombay phenotype is characterized by mutations or deficiency in Fucosyl transferase. There is a para-Bombay phenotype (denoted Ah, Bh, and ABh). These are observed in individuals with weakly expressed A or B, but not H, on their red cells; no A or B antigens are found in the plasma [Schroeder & Rayner (28), Mohn et al. (23)]. H transferase is not detectable, but appropriate A and B transferases are present. It is thought that the small amount of H substance synthesized is transformed completely to A or B by the respective transferase [Watkins (32)].

Family studies have shown that the Bombay phenotype (Oh) is due to the presence in homozygous state of a rare recessive gene [Yunis et al. (34)]. Because both parents must carry this recessive allele to transmit this blood type to their children, the condition mainly occurs in small closed-off communities where the recessive gene has a chance to find two parents with the same blood type [Balgir (5); Balgir (3); Bhatia & Sanghvi (8); Sringer et al. (30); Yunis et al. (34); De Zoyas (14)]. In the present case, we could not detect the Bombay Blood Group of the patient’s parents as they were not alive. His younger brother was found to be Bombay phenotype negative.

H-deficient Bombay phenotype is rare, since it occurs in about 1 in 10,000 individuals in India and 1 per 1,000,000 individuals in Europe [Oriol et al. (25)]. After the first report of Bombay Blood Group (Oh phenotype) from Mumbai (formerly Bombay) in 1952 by Bhende (11), several other workers detected this peculiar phenotype in India [Simmons & D’senna (29); Roy et al. (26)] and also in the European countries [Alosia et al. (1); Aust et al. (2)].

Later on, it was found that many of the European cases, which were initially labeled as typical Bombay phenotypes, turned out to be para-Bombay phenotypes after absorption-elution studies [Levine et al. (22); Lanset et al. (20); Bhatia & Solomon (10)]. Balgir (5) has shown 1 in 278 incidence of the Bombay phenotype among the Bhuyan tribal population of Orissa (India). Balgir (3) has also reported an incidence of 1 in 33 among the Kutia Kondh primitive tribe from Kandhamal district of Orissa. According to Balgir (5) the practice of endogamy and consanguinity amongst the Bhuyan tribal population and Kutia Kondh primitive tribe are the major factors for the relatively high prevalence of recessive rare alleles like Bombay Phenotype. Bhatia and Sanghvi (8) calculated the incidence of this phenotype as 1 in 13,000 individuals in Mumbai. Later on Bhatia and Sathe (9) found an incidence of 1 in 7600 after screening a large number of samples in Mumbai. Gorakshakar et al. (18) after systematic screening of the rural population from Ratnagiri and Sindhudurg districts of Maharashtra (India), reported the incidence of the Bombay phenotype as 1 in 4500 in that region.

Regarding the distribution and spread of the Bombay phenotype in different states of India, it is apparent that the phenotype is more common in the states of Western and Southern parts of India as compared to other states [Balgir (5)]. Of the 179 cases recorded by Sathe et al. (27), 112 (62.6%) cases belonged to the state of Maharashtra. A slightly higher frequency of the Bombay phenotype was also found in the

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neighboring state of Karnataka (14 cases), Andhra Pradesh (8 cases), Goa (6 cases), Gujarat (5 cases), Uttar Pradesh (5 cases), and so on in the decreasing order. There is no published data available in the literature on the caste/tribe-wise distribution of the Bombay phenotype in India. Moreover, most of the reported cases were either referred cases or were hospital cases seeking blood transfusions. Hence, the exact prevalence of the Bombay phenotype, which is based on random population screening (not based on hospital data), is not yet precisely known in India [Balgir (5)]. Further, based on the available information in India, it is interesting to note that the incidence of the Bombay phenotype is high in those states of India where consanguineous marriages are more prevalent (i.e., Andhra Pradesh, Tamil Nadu, Karnataka, Maharashtra, Gujarat, etc.) than in the other states [Balgir (5)].

The present case is a Muslim patient, and it is seen that endogamy and consanguinity are strictly followed in some Muslim families – especially in Bohra Muslims.

The Bombay phenotypes were also detected in Japan [Kaneko et al. (19); Okubo (24)], Malaysia [Sathe et al. (27)], Thailand [Sringarm et al. (30)], and Sri Lanka [De Zoyza (14)]. Yunis et al. (34) found seven individuals of Oh phenotype in two generations of an Indian family settled in the USA. They were natives of Orissa state. Similarly, according to Sathe et al. (27), 24 cases of Oh phenotypes were found in 11 unrelated Indian families settled in Natal, South Africa. Its incidence was 1 in 18,404. Most of these families were either Tamil or Telugu speaking. Therefore, their origin is presumed to be Andhra Pradesh or Tamil Nadu.

A large series of H-deficient individuals - Oh phenotypes (~1:1000) were found in a small French island 800 Km east of Madagascar in the Indian Ocean, called Reunion Island [Le Pendu et al (21)]. This indicates that the Bombay phenotype is mostly confined to South-East Asian countries [Balgir (5)]. In Japan, the incidence of Bombay and para-Bombay phenotypes was shown to be 1-2 in 300,000 [Kaneko et al (19)]. In Taiwan, para-Bombay phenotype has a frequency of 1:8000 [Yu et al (33)].

This group would be categorized as the O group because it wouldn’t show any reaction to anti-A and anti-B antibodies just like a normal the O group. When a cross matching with O group is done, then it would show cross-reactivity or incompatibility. Therefore reverse grouping or serum grouping has to be performed to detect this group.

Since the Bombay Blood Group is the rarest blood group, it is desirable to develop cryopreservation facilities for rare donor units. Every blood bank can easily maintain a rare blood type donor file from amongst their regular voluntary donors. If the blood banks can borrow or exchange rare blood units in times of need, a lot of problems related to rare blood groups like the Bombay Blood Group can be solved. This is only possible if each blood bank has a large number of committed regular voluntary donors.

**REFERENCES**


ABOUT THE AUTHOR

Mansoor Quli Khan, MBBS, MD, Ph.D. serves as a Senior Consultant Clinical Haemat-oncologist at Hosmat Hospital and other hospitals in Bangalore, India. He is a Faculty Member at Akamai University and was formerly the lead Consultant Haematoatologist and Head of the Haematology Section of the Central Hospital and Medical Laboratory, Riyadh medical Complex (MOH), Riyadh, Saudi Arabia.

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