

Identification of a rare blood group, “Bombay (Oh) phenotype,” in Bhuyan tribe of Northwestern Orissa, India

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BACKGROUND: Blood group serology plays a vital role in transfusion medicine. The Bombay (Oh) phenotype is characterized by the absence of A, B, and H antigens on red cells and occurs rarely, especially in tribal populations of India.

AIMS AND OBJECTIVES: This is a field-based random population study in the Bhuyan tribal community. The study reports three cases of the rare Bombay (Oh) phenotype for the first time in the Bhuyan tribe of Sundargarh district in North-Western Orissa.

MATERIALS AND METHODS: Taking informed consent, red blood cells of 836 Bhuyan subjects were tested with three antisera, i.e., anti-A, anti-B, and anti-H (lectin) for forward reaction. Agglutinations of plasma with A, B, and O (H) red cells (reverse reaction) were also tested for the presence or absence of antibodies in the serum. Specialized tests like absorption-elution, titration of naturally occurring antibodies at different temperatures, inhibition of anti-H by O saliva secretor, and determination of secretor status were performed.

RESULTS: Three cases of a rare blood group, Bombay (Oh) phenotype, (2 out of 244 Khandayat Bhuyan and 1 out of 379 Paudi Bhuyan from Hemgiri and Lahunipara blocks, respectively) in the Bhuyan tribe of Sundargarh district in North-Western Orissa were detected, giving an incidence of 1 in 122 in Khandayat Bhuyan and 1 in 379 in Paudi Bhuyan, with an average of 1 in 278 among the Bhuyan tribal population. This incidence is high in comparison to earlier studies reported from India.

CONCLUSIONS: The practice of tribal and territorial endogamy in a smaller effective populations (for example, there are only 3,521 individuals in Paudi Bhuyan) results in smaller marital distance and inbreeding, leading to increased homozygous expression of rare recessive genetic characters like the Bombay (Oh) phenotype. This study further testifies that the incidence is higher in those states of India where the consanguinity is a common practice.

Key words: Blood groups, Bombay phenotype, Khandayat Bhuyan, Paudi Bhuyan, Primitive tribe

devoid of the H antigen on red cells in Bombay (India) who had antibodies in plasma reacting with all the red cells exhibiting the normal red cell ABO phenotypes.^[1] This phenotype was characterized by the absence of A, B, and H antigens on red cells, and the serum of these persons had anti-A, anti-B, and anti-H, reacting with all O group bloods. 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+). Watkins and Morgan^[2] and Gerard *et al.*^[3] 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.^[4,5]

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.^[5] More recently, a large series (42 H-deficients) of H-deficient individuals (~1:1000) were found in a small French island 800 km east of Madagascar in the Indian Ocean, called Reunion Island.^[6] The two phenotypes resulted from products, or lack of products, of two different alleles of *FUT1* and *FUT2* genes;^[6] the same and also additional alleles of both *FUT1* and *FUT2* were documented in other populations, particularly in Japan, where the incidence of Bombay and para-Bombay individuals was shown to be 1-2 in 300,000.^[4] In Taiwan, para-Bombay phenotype has a frequency of 1:8000.^[7] In India, the *FUT1* mutation travels almost always (one exception) with a total deletion of *FUT2*; in Reunion Island (Caucasian), the major inactivating mutation of *FUT1* travels almost always with the inactivating mutation of *FUT2*; and the main Oriental inactivating mutations of *FUT1* travel almost always with the wild-type *FUT2*.^[8]

Introduction

The existence of a human H/h genetic polymorphism was first established by the discovery of an individual

After the first report of Oh phenotype from Mumbai (formerly Bombay) in 1952 by Bhende *et al.*,^[1] several other workers detected this peculiar phenotype in India^[9,10] and also in the European countries.^[11,12] 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.^[13-15] The difficulty with the Bombay phenotype is that the individuals having blood group of Bombay phenotype (Oh) 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.

The present study, being a part of the screening for hereditary hemolytic anemia (i.e., hemoglobinopathies and allied disorders) in the tribal communities, was designed with the following aim and objective: To study and identify the rare blood groups like Bombay (Oh) phenotype detected, if any, in the Bhuyan tribe of North-Western Orissa.

Materials and Methods

A total of 244 Khandayat Bhuyan from Ratansara village in Hemgiri Block and 379 Paudi Bhuyan tribals from three villages, namely, Badjal, Budhabhuin, and Kuliposh colony in Lahunipara Block of Sundargarh district in Northwestern Orissa were screened for hemoglobin disorders and ABO and Rh (D) blood groups during the year 2003-04. From these villages, venous blood samples (1-2 ml in EDTA containing vials) and saliva samples (in tubes) were collected. Members of both the genders were included in the study and prior written informed consent was obtained from each subject. The Human Ethical Committee of our Institute had approved the screening and intervention for hereditary hemolytic disorders among the major tribal communities of Sundargarh district in Orissa. These samples were transported under ice-cold conditions to the laboratory within 24 h of collection and were analyzed using tube technique as per the instructions of the manufacturer of the anti-sera (Tulip Diagnostics Private Limited, Panaji, Goa, India).

The ABH phenotyping was carried out by applying the standard forward and reverse grouping tube techniques.

The Bombay phenotype detected was further confirmed by certain specialized tests like absorption-elution studies, titration of naturally occurring antibodies at different temperatures, hemagglutination-inhibition study on anti-H by O saliva secretor, and secretor-status of the person as described by Flynn^[16] and Boorman and coworkers.^[17]

Results

Two hundred and forty-four Khandayat Bhuyans from Ratansara village in Hemgiri Block and 379 Paudi Bhuyan tribals from Badjal and Budhabhuin villages and Kuliposh colony in Lahunipara Block of Sundargarh district were screened. Two unrelated women, aged 25 and 30 years, from the former Block and a 30-year-old woman from Kuliposh Colony of the latter Block showed no agglutination of red blood cells with anti-A, anti-B, and anti-H antisera, but the plasma of these individuals showed the presence of anti-A, anti-B, and anti-H antibodies, indicating the presence of the Bombay phenotype. All the three cases were non-secretors of ABH, i.e., Le (a+).

Discussion

The most striking finding of the present study is the detection of three unrelated cases of Bombay (Oh) phenotype in Paudi Bhuyan (a primitive tribe) and Khandayat Bhuyan from Lahunipara and Hemgiri Blocks, respectively, of Sundargarh district in North-Western Orissa, the Central-Eastern part of India. To the best of our knowledge, except in one study,^[18] no case of the Bombay phenotype has ever been reported among the primitive tribes from the state of Orissa. The present report deals with additional 3 cases of the rare Bombay phenotype.

The present study showed a 1 in 122 incidence of the Bombay phenotype among Khandayat Bhuyan tribe and 1 in 379 in Paudi Bhuyan, with an average of 1 in 278 among the Bhuyan tribal population of Orissa. Since the population size of Paudi Bhuyan tribe is relatively small (3,521 individuals as per Paudi Bhuyan Development Agency at Khuntagaon), and the

practice of endogamy is strictly followed, inbreeding and consanguinity amongst them is not ruled out, which may be one of the major factors for the relatively high prevalence of recessive rare alleles like Bombay phenotype among the Khandayat and Paudi Bhuyan tribe. Balgir^[18] has reported an incidence of 1 in 33 among the Kutia Kondh primitive tribe from Kandhamal district of Orissa. Bhatia and Sanghvi^[19] calculated the incidence of this phenotype as 1 in 13,000 individuals in Mumbai. Later on, Bhatia and Sathe^[20] found an incidence of 1 in 7600 after screening a large number of samples in Mumbai. Gorakshakar *et al.*,^[21] after systematic screening of the rural population from Ratnagiri and Sindhudurg districts of Maharashtra, reported the incidence of the Bombay phenotype as 1 in 4500 in that region, while Moores^[22] reported its incidence as 1 in 18,404 amongst Indians settled in South Africa.

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. Of the 179 cases recorded by Sathe *et al.*,^[22] 112 (62.6%) cases belonged to the state of Maharashtra. A slightly higher frequency of the Bombay phenotype was also found in the 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 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. 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.

The Bombay phenotypes were also detected in Japan,^[4,23] Malayasia,^[22] Thailand^[24] and Sri Lanka.^[25] Yunis *et al.*^[26] found seven individuals of Oh phenotype in two generations of an Indian family settled in the USA.

They were natives of Orissa state. Similarly, Moores^[22] found 24 cases of Oh phenotypes in 11 unrelated Indian families settled in Natal, South Africa. Most of these families were either Tamil or Telugu speaking. Therefore, their origin is presumed to be Andhra Pradesh or Tamil Nadu. More recently, a large series of H-deficient individuals (~1:1000) were found in Reunion Island in the Indian Ocean.^[6] This indicates that the Bombay phenotype is mostly confined to South-East Asian countries.

It is suggested that further molecular research on the Bombay phenotype is required, which may add to the existing variant phenotypes and elucidate the evolutionary significance and the operation of natural selection among the Khandayat and Paudi Bhuyan primitive tribes of India.

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Appendix

The Bhuyan tribe is an endogamous community, which is mostly confined to Sundargarh district of Orissa. This is a border district, with Jharkhand state in the North, Chhattisgarh state in the West, Keonjhar district in the East, and Jharsuguda, Sambalpur, and Deogarh districts in the South [Figure 1].

However, there are sporadic families of the Bhuyan tribe seen in Anugul, Deogarh, Jharsuguda, and Keonjhar districts. Some of their families have also migrated to Assam to work in the tea plantations as labor force.^[27-29] The Bhuyan tribe, although originally of one ethnic stock, is now divided into three social groups namely, the Hill Bhuyan (Pahari or Paudi Bhuyan), Paraja Bhuyan (Common People), and the Paik or Khandayat Bhuyan (Warrior), which are distinguished from each other on the basis of three grades of primitive culture in the state



Figure 1: Map of Orissa showing thirty districts and study area (shaded)

of Orissa. The Hill Bhuyan, the primitive and backward section, represents the hunting and food gathering stage of economic life; they also practice a rudimentary shifting cultivation and have a primitive culture. The Paraja Bhuyan section represents a more advanced culture, and practise plough-cultivation and food production. The Khandayat Bhuyans have the most advanced culture,

which equates them with other nontribal populations of the region. Inter-group marriages do not take place at all. Reproductively and genetically, they are completely isolated from each other.

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