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Genetic Analysis of ABO and Rh Blood Groups in Backward Caste Population of Uttar Pradesh, India

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Abstract

A series of glycoproteins and glycolipids on red blood cell surface constitute blood group antigens. These are AB, A, B and O in ABO blood group system and Rh in rhesus blood group system. A total of 1065 unrelated Backward Caste (OBC) individuals from Uttar Pradesh were studied for the phenotype and allele frequency distribution of ABO and Rh (D) blood groups. Total 1065 samples analyzed, phenotype B blood type has the highest frequency 36.81% (n=392), followed by O (32.68%; n=348), A (23.66%; n=252) and AB (6.85%; n=73). The overall phenotypic frequencies of ABO blood groups were B>O>A>AB. The allelic frequencies of O, A, and B alleles were 0.5819, 0.1674 and 0.2506 respectively. Out of total 1065 samples, 1018 (95.59%) samples were Rh-positive and 47 (4.41%) were Rh-negative. Phenotypic frequency of Rh-negative in Koari, Yadav, Kurmi and Maurya samples were 0.99%, 4%, 1.4% and 7.6% respectively.

Keywords: ABO blood groups, antigen, allele frequency, Backward Caste

Introduction

The ABO blood group system is the most clinically important blood group system because antibodies against A or B or both antigens are naturally present in the serum of persons whose red cells express blood group B, A, or O. The ABO incompatible transfusions are potentially fatal. It follows that universal blood typing with DNA-based methods alone cannot be considered in the absence of a totally robust method for predicting ABO phenotype. The molecular basis of the ABO blood group system was elucidated in 1900. Landsteiner (1900) has discovered three different blood types (A, B, and O) of this ABO blood group system. The fourth blood type (AB) was discovered by DesCasterllo and Sturli (1902). The ABO antigens were originally found on red cells (Landsteiner, 1990), but later they were also found on the surface of various types of cells as well as in secretions (Davidson and Stejskal, 1962).

The ABO locus is located on chromosome 9 at 9p34.1-q34.2 and encodes glycosyltransferases. ABO locus has three main allelic forms: A, B and O (Larsen *et al*., 1990; Hasoi, 2008; Watkins, 1980; Yamamoto *et al*., 1995) and Yamamoto *et al*. (1990) cloned and determined the structure of the gene. *ABO* gene spans about 18-20 kilobases (kb) organised into seven exons. Exons 6 and 7 contain 77% of the full coding region and encode the domain responsible for catalytic activity (Daniels, 2009). Exon 7 contains most of the largest coding sequence. Exon 6 contains the deletion found in most O alleles. The exons range in size from 28 to 691 bp (Yamamoto *et al*., 1990). The *ABO* gene codes for the glycosyltransferase that transfer specific sugar residues to H substance, resulting in the formation of group A and B antigens. A and B alleles have seven nucleotide substitutions. Four nucleotide substitutions are translated into different amino acid substitution. It has made it possible to analyze genetically ABO blood group antigens using molecular biology techniques (Larsen *et al*., 1990; Hasoi *et al*., 1998; Ogasaware *et al*., 1996, 1998; Yamamoto *et al*., 1990). The antigens A, B, and their variants result from functional glycosyltransferase genes capable of transferring N-acetyl-D-galactosamine or D-galactose or both to the nonreducing ends of suitable oligosaccharide chains found on red cell membrane glycoproteins and glycolipids. The red cell phenotype denoted O occurs because the glycosyltransferase gene that generates A or B or both antigens is inactive (Anstee, 2009).

The ABO blood group system was the first genetic polymorphism defined in human beings. Since that time the blood groups has played a prominent role in the study of human polymorphisms, and because of its easy classification into different phenotypes, relatively simple mode of inheritance, and different frequencies in different populations, blood groups are useful genetic markers in family, and population studies, and in linkage analysis (Ali *et al*., 2005). Its distribution is studied by several scientists in almost all the races and populations of the world like-Nigeria (Ahmad and Obi, 1998; Adeyemo and Soboyejo, 2006; Enosolease and Bazuaye, 2008; Gaertner *et al*., 1994; Jeremiaha, 2006), Kenya (Lyko *et al*., 1992), Palestine (Alishtayeh *et al*., 1988), Iraq (Mohamad, 2010), Sudan (Kalmokova and Konova, 1999; Hassan, 2010), Pakistan (Afzal *et al*., 1977; Ali *et al*., 2005; Anees and Mirza, 2005; Bhalti, 1998; Hameed *et al*., 2002; Khaliq *et al*., 1984; Khurshid *et al*., 1992; Khan *et al*., 2004; Mian and

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Farooq, 1999; Shamim *et al*., 2002; Yousuf *et al*., 1988), Bangladesh (Talukder and Das, 2010) and Saudi Arab (Al-Himaidi and Umar, 2002; Abdullah, 2010; Sarhan *et al*., 2009), Jordan (Hanania *et al*., 2007), Iran (Boskabady *et al*., 2005), Nepal (Pramanik and Pramanik, 2000), India (Chakraborty, 2010; Deepa *et al*., 2011; Majumdar *et al*., 1992; Rai and Kumar, 2010) .

The Rh blood group, popularly referred to as Rhesus, is second only to the ABO system in its importance in transfusion medicine. Although the Rh system is highly polymorphic, and comprises at least 44 distinct antigens, clinically the most significant polymorphism is due to the presence or absence of the Rh (D) antigen on red cells. The Rh antigens are carried on three nonglycosylated transmembrane proteins that are encoded by two genes, *RHD* and RHCE (Arce *et al*., 1993; Avent *et al*., 1991; Simse *et al*., 1994). Alternative mRNA splicing is responsible for the production of two distinct polypeptides from the single *RHCE* gene. Lack of D antigen expression is usually due to the absence of the entire *RHD* gene from the genome of Rh (D) negative individuals (Colin *et al*., 1991).

Approximately 300 different types of blood groups are identified so far, indeed, the Rh and ABO antigens are still the clinically most significant (Klein and Anstee, 2005) and genetically most polymorphic of all human blood group systems to date (Blumenfeld and Patnaik, 2004). However, ABO are carbohydrate antigens (Watkins, 1966) depending on the enzymatic activity and specificity of allelic glycosyltransferases (Yamamoto *et al*., 1990), whereas Rh antigens are protein motifs (Gahmberg, 1982; Moore *et al*., 1982), whose surface expression entails an interaction of two genetic loci (Huang *et al*., 2000; Le *et al*., 2006). The protein nature endows Rh antigens, particularly the more recently evolved D antigen, with the inherent ability to mount potent alloimmune reactions to counteract such conflicting situations as fetal-maternal incompatibility (Huang and Ye, 2010). The present study was carried out to determine the ABO and Rh blood groups frequencies in OBC population of UP.

Material and methods

Blood samples were taken by finger pricks from 1065 unrelated individuals of both sexes of OBC population, and open slide method of ABO blood groups testing was followed (Bhasin and Chahal, 1996). ABO Typing antisera of Span were used for ABO Typing. The gene frequencies for both the systems were calculated according to the method of Mourant *et al*. (1976). The details of each subject such as name, age, sex etc. were collected using a brief questionnaire. Informed consent was taken from each subject. Samples were collected from Koari, Yadav, Kurmi and Maurya population of Uttar Pradesh.

Results and discussion

In total 1065 samples analyzed, phenotype B blood type has the highest frequency 36.81% (n=392), followed by O (32.68%; n=348), A (23.66%; n=252) and AB (6.85%; n=73) (Tab. 1). The overall phenotypic frequencies of ABO blood groups were B>O>A>AB (Fig. 1). The allelic frequencies of O, A, and B alleles were 0.5819, 0.1674 and 0.2506 respectively (Fig. 2). Total number of samples were also categorized by gender, 406 samples were

Tab. 1. Distribution of the ABO blood group and their allele frequencies among OBC Population

Phenotype	Observed Number	Percentage	Expected Number	Allele frequency
O	348	32.676	360.6	0.5819
A	252	23.66	237.3	0.1674
B	392	36.81	377.5	0.2506
AB	73	6.85	89.4	

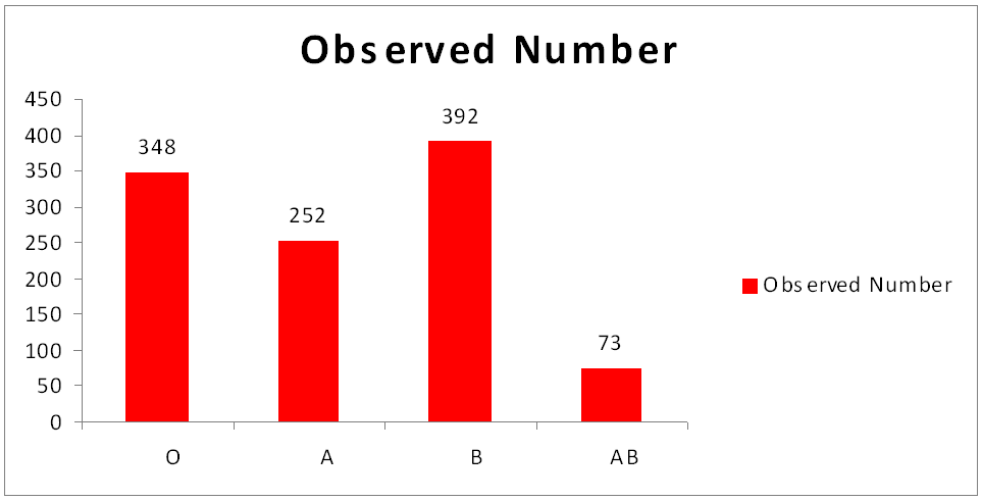

Fig. 1. Phenotypic number of different ABO groups observed

Tab. 2. Total number of samples classified according to gender

Sections	No. of O phenotype	No. of A phenotype	No. of B phenotype	No. of AB phenotype	Total
Females	141	100	135	30	406
Males	207	152	257	43	659
Total	348	252	392	73	1065

Tab. 3. Sub-caste wise distribution of ABO blood group among OBC population

Sections	O phenotype		A phenotype		B phenotype		AB Phenotype		Total
	No.	%	No.	%	No.	%	No.	%	
Koari	70	34.5	50	24.6	67	33	16	7.9	203
Yadav	80	40	34	17	73	35.5	13	6.5	200
Kurmi	57	26.89	54	25.47	72	33.96	29	13.68	212
Maurya	141	31.33	114	25.33	180	40	15	3.33	450
Total	348	32.68	252	23.66	392	36.81	73	6.85	1065

Tab. 4. Gender wise distribution of ABO blood group among Koari population

Sections	No. of O phenotype	No. of A phenotype	No. of B phenotype	No. of AB phenotype	Total
Females	41	30	37	10	118
Males	29	20	30	06	85
Total	70	50	67	16	203

of females and 659 of males (Tab. 2). In female samples 141 individuals had O blood group, 100 individuals had A blood group, 135 individuals had B blood group and 30 individuals had AB group. In 659 male samples, O, A, B and AB blood groups were found in 207, 152, 257 and 43 individuals respectively. In female samples the phenotypic frequencies were O>B>A>AB, whereas in male samples overall phenotypic frequencies were B>O>A>AB (Tab. 2). The variation in phenotypic frequencies between male and female might be due to small sample size of male sample.

Tab. 5. Gender wise distribution of ABO blood group among Yadav population

Sections	No. of O phenotype	No. of A phenotype	No. of B phenotype	No. of AB phenotype	Total
Females	25	7	13	4	49
Males	55	27	60	9	151
Total	80	34	73	13	200

Samples were also categorized on the basis of subcaste. 203 samples were of Koari, 200 samples were of Yadav, 212 samples were of Kurmi and 450 samples were of Maurya subcaste. The highest frequency in Koari was of O type (34.5%; n=70), followed by B (33%; n= 67), A (24.6%; n=50) and AB (7.9%; n=16) (Tab. 3). In female samples the phenotypic frequencies were O>B>A>AB, whereas in male samples overall phenotypic frequencies were B>O>A>AB (Tab. 4). In 200 Yadav samples the highest frequency was of O type (40%; n=80), followed by B (35.5%; n= 73), A (17%; n=34) and AB (6.5%; n=13)(Tab. 3). In Yadav females the highest frequency was of O type (51%), followed by B (26.53%), A (14.23%) and AB (8.16%). In Yadav males the highest frequency was B (39.73%) followed by O (36.42%), A (17.88%) and AB (5.96%) (Tab. 5). Whereas in 200 Kurmi samples the phenotypic frequencies of O, A, B and AB were 26.89% (n=57), 25.47% (n=54), 33.94% (n=72) and 13.68% (n=29) respectively (Tab. 3). 109 samples were of females and 103 samples were of males. The phenotypic frequency in females were B>A>O>AB and in males phenotypic frequencies were

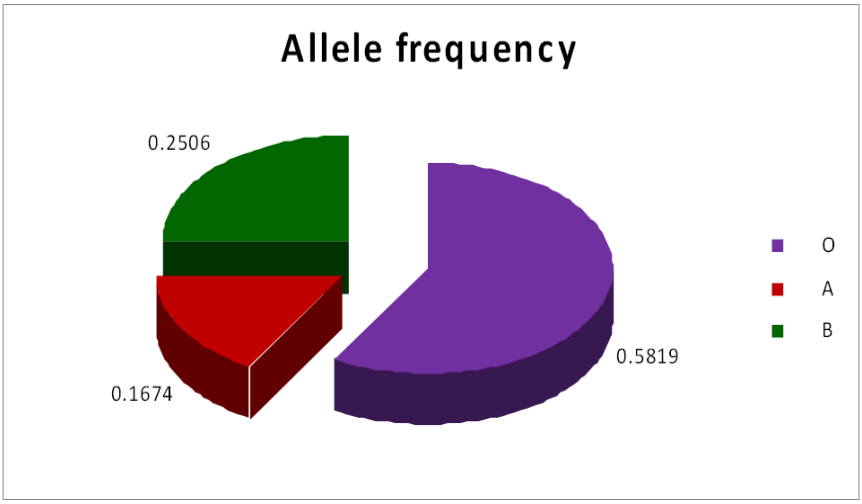

Fig. 2. Allelic frequencies of A, B, and O blood groups in total 1065 samples analyzed

Tab. 6. Gender wise distribution of ABO blood group among Kurmi population

Sections	No. of O phenotype	No. of A phenotype	No. of B phenotype	No. of AB phenotype	Total
Females	28	30	38	13	109
Males	29	24	34	16	103
Total	57	54	72	29	212

Tab. 7. Gender wise distribution of ABO blood group among Maurya population

Sections	No. of O phenotype	No. of A phenotype	No. of B phenotype	No. of AB phenotype	Total
Females	47	33	47	03	130
Males	94	81	133	12	320
Total	141	114	180	15	450

Tab. 8. Rh blood group among OBC population

Phenotypes	Observed		Allele frequencies
	Number	Percentile	
Rh(Anti D)+	1018	0.9559	D=0.7899
Rh(Anti D)-	47	0.0441	d=0.2101
Total	1065	1.0000	

B>O>A>AB (Tab. 6). The highest frequency in Maurya samples was of B type (40%; n=180), followed by O (31.33%; n= 141), A (25.33%; n=114) and AB (3.33%; n=15)(Tab. 3). In female samples the phenotypic frequencies were O>B>A>AB, whereas in male samples overall phenotypic frequencies were B>O>A>AB (Tab. 7) (Fig. 3). Out of total 1065 samples, 1018 (95.59%) samples were Rh-positive and 47 (4.41%) were Rh-negative (Tab. 8). Phenotypic frequency of Rh-negative in Koari, Yadav, Kurmi and Maurya samples were 0.99%, 4%, 1.4% and 7.6% respectively (Tab. 9).

Although several reports are published on RBC antigens of various castes from U.P. as well as from all over the India like- Brahmins (Guniyal, 2006; Mukhopadhyay and Kshatriya, 2004; Tewari and Bhasin, 1968), Rajputs (Chaudhary and Malik, 1997; Kumar *et al*., 2009a; Mukhopadhyay and Kshastriya, 2004; Pattanayak, 2006; Warghat *et al*., 2011), Scheduled Caste population (Kushwaha *et al*., 1990; Mandal, 1992; Patni and Yadav, 2003; Rai *et al*., 2009a, b; Sidhu, 2003; Thukral and Bhasin, 1990), muslim population (Ara *et al*., 2008; Kumar *et al*., 2010; Rai *et al*., 2010; Srivastava, 1975) but very few reports are available about ABO distribution in Backward castes population (Kumar *et al*., 2008, 2009b; Prabhakar *et al*., 2005; Reddy and Reddy, 2005). In all earlier published reports on ABO distribution in OBC population, the sample size was very small and in present study the sample size is comparably large

Tab. 9. Rh blood group among OBC population

Caste	Total number	Rh D Positive			Rh D Negative		
		Observed Number	Percentage	Allele Frequency	Observed Number	Percentage	Allele Frequency
Koari	203	201	99.01	0.901	2	0.99	0.099
Yadav	200	192	96	0.8	8	4	0.2
Kurmi	212	209	98.58	0.8808	3	1.42	0.1192
Maurya	450	416	92.4	0.7250	34	7.6	0.2750
	1065	1018			47		

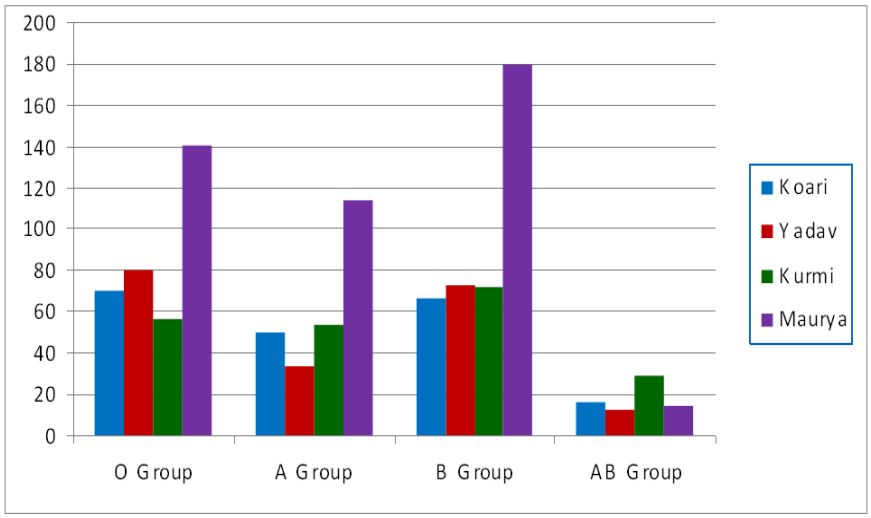

Fig. 3. Gender-wise categorization of different ABO blood groups phenotypes

The frequency of ABO blood group varies from race to race. The allelic frequencies of the total population of the world is found to be O=62.3%; A=21.5% and B=16.2%. Among Western European, 42% have group A, 9% group B, 3% group AB and remaining 46% group O. American blacks generally have frequencies of A, B, AB and O blood groups of 27%, 20%, 4%, and 49% respectively (Conteras and Lubenko, 2001). Among the population of Southwest Asian countries (Saudi Arabia, Jordan, Kuwait, Syria, Iraq, Iran) the frequencies of alleles A and B are about 23 and 15 respectively (A>B), except in Afghanistan, where the allele B is higher than O and A (Ara *et al*., 2008; Mourant *et al*., 1976; Tills *et al*., 1983). Our frequencies are comparable with the neighboring country Afganistan and Pakistan, where the highest phenotypic frequency was of B group.

The Rh distribution also varies within any group of population. The recessive allele (d)ranges from as high as 40% to its virtual absence in Chinese Australian aborigines, Negrito etc. Exceptionally high incidence of Rh negatives yielding frequency of recessive allele (d) in the range of 50 to 60% have been reported in Basque (Europe) and Berbers of Moracco (Mourant *et al*., 1976). Rh -negative blood group is documented as 5.5% in south India, 5% in Nairobi, 4.5% in Nigeria, 7.3% in Lahore, 7.7% in Rawalpindi (Bhalti and Amin, 1996; Das *et al*., 2001; Mawuagi, 1999; Majeed and Haye, 2002). In present study, the percentage of Rh-negative was found to be 4.41% which was comparable to the earlier reports. Over the years, the Rh blood group system has been distributed among any population to keep the frequency of Rh-negative very low since clinical situations could arise through Rh incompatibility.

The need for blood group prevalence studies is multipurpose, as besides their importance in evolution; their relation to disease and environment is being increasingly sought in modern medicine. The ABO blood group has been reported to be associated with many diseases like-cancer (Dabelsteen and Gao, 2005; Guleri *et al.*, 2005; Iodice *et al.*, 2010; Nozoe *et al.*, 2004; Sharma *et al.*, 2007; Vadivelu *et al.*, 2004; Xie *et al.*, 2010), eye diseases (Dhillon and Shergill, 2004; Khan *et al*., 2009; Mourant *et al*., 1976; Zaree *et al*., 2006), skin diseases (Gangopadhyay *et al*., 2006; Valikhani *et al*., 2007), cardiovascular diseases (Biswas *et al.*, 2008; Skaik, 2009), diabetes (Koley, 2008; Okon *et al*., 2008), malaria (Deepa *et al*., 2011; Jeremiaha *et al*., 2010), infectious diseases like-Smallpox (Krieger and Vilente, 1969), Leprosy and cholera (Urade and Chakravarty, 1999) and Cholera (Harris *et al*., 2005) though the explanation for the association between ABO blood groups and disease is still unclear. In addition information of blood groups is very useful in blood transfusion and organ transplantation medicine, in human population migration and evolution study, in genetic research and in parental dispute cases.It is, therefore, imperative to have information on the distribution of these blood groups in any population group. The present study is therefore, useful in providing information on the status of ABO blood group distribution in the OBC population of Uttar Pradesh.

Conclusions

The frequency of ABO and Rh phenotypes in OBC appears to be similar to Asian data. The study results show that the most frequent blood group in the OBC caste group of UP is group B and the rarest is group AB and Rh-negative frequency is 4.41%. This study has a significant implication regarding the management of blood banks and transfusion services in this area. Knowledge of blood group phenotype distribution is also important for clinical studies (for example disease association), as well as for population studies. It is necessary to conduct similar well designed studies in other states of India in order to determine the blood group frequencies in them. The data generated in the present study and several other studies of different geographical region of India may be useful for health planners, while making efforts to face the future health challenges in the region. In short, generation of a simple database of blood groups, not only provides data about the availability of human blood in case of regional calamities, but also serves to enable insight into possibilities of future burden of diseases.

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