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Research Article

Distribution of ABO and Rh (D) allele frequency among the populations of Yilmana Denssa and Mecha, Ethiopia

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ABSTRACT

Background: The distributions of ABO blood group and Rh (D) factor is important for population distribution and genetics studies in multi-ethnic countries. Such studies are not completely covered the parts of Ethiopia, a country with multi ethnic groups. Therefore, this study is designed to decipher the population blood group distribution frequency in the population of West Gojam.
Methods: A total of 200 Volunteer individuals were participated in this study as a study subject to determine blood group in the study population by the antigen-antibody agglutination test. The results of tests were subjected to allelic frequency distribution calculation using Panmictic index.
Results: The total of the two sites (Yilmana Denssa and Mecha) had O (r=0.624), A (p=0.2299) and B (q=0.1456) of allele frequency and the genotypic frequency were AA (p2=0.05285), AO (2pr=0.287), BB (q2=0.0212), BO (2qr=0.1818), AB (2pq=0.0669) and OO (r2=0.39). Out of the tested samples 91%, was recorded Rh+ in the same population, with frequency of 0.7 for allele D confirming the majority of the population in this area as Rh+. Fixation indices (F) for ABO gene were 17 and 22 Yilmana Denssa and Mecha respectively which was higher in Ylmana Denssa.
Conclusions: The Panmictic index (P)was 78 lower than Mecha site for ABO gene and the observed heterozygosity (Ho) for ABO gene was 0.53 indicating large genetic variations in populations of the study area. Therefore, the findings of this study can be used as a baseline for the genetic diversity of the study area as a reference for Ethiopian ABO blood group phenotypes and genotypes studies.

Keywords: ABO blood, allele frequency, Rh factor

INTRODUCTION

The ABO and Rhesus (Rh) blood groups are the most important in clinical aspects. The differential distributions of these blood groups have been observed in various populations all over the world. The frequencies show considerable variation in different geographic locations reflecting the genetic and ethnic diversity of human populations.[1] All human populations share the same blood group systems inherited from common ancestor but they differ by their frequencies. Allele frequency is the proportion of all copies of a gene that is made up of a particular gene variant (allele). It is also the relative proportion of all alleles of a gene that are of a designated type.[2] In population genetics, allele frequencies are used to depict the amount of genetic diversity at the individual, population, and species level. The ABO blood system shows more than one type of dominance in the alleles (combination of complete dominance of A and B over O and co-dominance of A and B) and contains four types of phenotypes with six genotypes types (phenotype A with the genotype AA and AO, phenotype B with the genotype BB and BO, phenotype AB with the genotype AB and phenotype O with the genotype OO). The genotypic and phenotypic frequencies of a given locus with certain number of alleles depend on the frequency of the alleles and the mating system of a population.[3-4] Genetic marker for

identity testing and paternity analysis are also dependent on known allele frequencies in the populations.[5] Genes exist in a number of different forms (alleles) and can undergo mutation. Alleles commonly exist in two (diploid), three (triploid) or four (tetraploid) forms of a gene that can have the same place on homologous chromosomes and are responsible for alternative traits.

For the A and B antigens, the primary gene products are carbohydrate, not protein antigens and synthesized by a series of enzymatic reactions catalysed by enzymes glycosyl transferases. The final step of their biosynthesis is catalyzed by A and B transferases encoded by the functional A and Balleles at the ABO genetic locus, respectively. Their allele frequencies vary among different races, which furnish interesting questions in population studies, anthropology, and human genetics. A and B antigens are not restricted to humans. The identical or similar antigens have been identified in other species. Similarly, glycosyl transferases other than A and B transferases exhibit similar specificity in reactions. This also creates evolutionary and enzymological interest in studying ABO alleles and their biosynthesis since these antigen expression exhibits dynamic changes during development and pathogenesis.[6]

The Rh blood system is important since Rh antibody causes severe hemolytic disease of the newborn (HDN) and their importance in blood transfusion.[7] It is clinically the most important protein-based blood group system with 49 antigens so far described. The antigens are located on two Rh proteins: RhD and RhCE and are produced by differences in their protein sequences. In CD nomenclature, they are termed CD240D and CD240CE. Unlike proteins of other blood groups, Rh proteins are expressed only in the membranes of red blood cells and their immediate precursors. The Rh is genetically complex but it is simply described in terms of a single pair of alleles, D and d. Rhesus positive (Rh+ve) persons are DD and Dd whereas, Rhesus negative (Rh-ve) are dd.[8] Different studies have reported the protective role of blood groups from severe malaria as a genetic selection mechanism in some parts of Ethiopia which is also common in other malaria endemic countries of Africa.[9-11] In Ethiopia, recent studies have reported high frequency of O blood group among population with diverse ethnic background which is not representative enough to judge the entire population allelic distribution. Only a few studies have investigated blood groups in Ethiopia.[9,10,13]

There is a limited information on the frequencies of ABO and Rh (D) blood groups in Ethiopian general population and particularly in the area of Yilmana Denssa and Mecha town.[10] Therefore, in this study, the distribution of ABO and Rh (D) allele frequencies among the populations of Yilmana Denssa and Mecha, Ethiopia were assessed to lay a base for population genetics studies.

METHODS

Description of the study site and type of study

Yilmana Denssa is located at 2000 m above sea level (ASL) at 11.4°N as well as 37.16°E whereas, Mecha is located at 2179 m ASL, 11.28°N and 37.49°E, respectively, in Amhara regional state, North West Ethiopia. Samples were collected from the two towns and a cross sectional descriptive study was conducted from December 2012 to January 2013.

Sample collection and blood test

A total of 200 blood samples were taken from both healthy and patient individuals, 100 samples at each Yilmana Denssa and Mecha health centers. About 7 to 10 ml blood samples were taken by an experienced laboratory technician in two separated 5 ml EDTA test tubes and invert for 8 to 10 times to mix with the anti-coagulant. The blood samples were centrifuged at 4000 to 5000 rpm for 25 min. The samples products (plasma 55%, Buffy coat < 1% and erythrocyte about 45% of whole blood) were separated. The antigen antibody agglutination test was done on slide. Anti A, anti B and anti D antibodies were dropped parallel to each other on the slide. The antigen- antibody agglutination reaction was observed visually and the results were recorded.[14]

Statistical analysis

The frequencies of O, A and B alleles belonging to ABO blood group system for the population were estimated from ABO blood group phenotype data.

a) Observed heterozygosity (Ho) Shannon diversity index for ABO gene was calculated on the basis of estimated allele frequencies as:

$$\mathrm{Ho} = 1 - \sum_{n=1}^{n} \mathrm{p2i}$$

Where pi^2 = genotype frequency of observed heterozygotes.

b) Expected heterozygosity (He) Hardy-Weinberg Equilibrium was calculated assuming the occurrence of all the three alleles of ABO gene in equal frequency (p = q = r = 1/k where k is the number of alleles) using:

$$\mathrm{He} = 1 - \sum_{n=1}^{n} \mathrm{p2i}$$

Where pi^2 = genotype frequency of expected homozygosity and He = 0.6666 for ABO gene considering 3 alleles.

c) Fixation index (F) for ABO gene in each population was estimated as:

$$F = \frac{He - Ho}{He}$$

F was expressed in percentage and Panmictic index (**P**) was calculated as: p = 1- F.

RESULTS

Table 1: Estimates of fixation (f) and panmictic (p) indexes and observed vs. expected for abo gene in the study area (N=200).

Population ABO phenotypes	Obs. H (Ho)	Exp. H (He)	X^2	
O	0.39	0.39	0	
A	0.34	0.34	0	
B	0.21	0.20	0.02	
AB	0.06	0.07	0.07	
Rh(D)+VE	0.91	0.7	0	
Rh(d)-VE	0.09	0.3	0	
Population	Obs. H (Ho)	Exp. H (He)	F	P
Yilmata Densa total (ABO) N=100	0.55	0.67	17.10	83
Mecha Total N=100 (ABO)	0.52	0.67	22.43	78
Grand total N=200	0.54	0.67	20	80

P= manmictic index; F= fixation index X^2=chi-square, Obs. H (Ho)=observed hetrozygosity; Exp. H (He) = Expected hetrozygosity

Among the samples collected from Mecha, 38% A, 36% O, 24% B and 2% were AB blood group. Regarding the Rh type the majority of the samples were Rh positive (92%) and the Rh negative were only 8%. The frequency of individual ABO groups studied in Yilmana Denssa were O (42%); A (30%), B (18%) and AB (10%). Similarly, as in Mecha area, the majority of the populations were Rh positive which accounts for 90%. Collectively at two sites 39% O, 34% A, 21% B and 6% AB blood groups were recorded. Allele frequencies showed a high frequency of the allele i over IB and IA alleles in the order of (i > IA > IB), respectively. Out of the 200 tested samples 91% were recorded to be Rh+ ve in the same population, with frequency of 0.7 for allele D. Relatively, the majority of the population in this area was Rh positive. Expected Frequency (genotypic frequency) was calculated from allele frequencies using $A = p^2+2pr$, $B = q^2+2qr$, $AB = 2pq$, $O = r^2$ to calculate the expected number and multiplied by the total samples. The allelic frequency of the three common alleles were O (r = 0.6), A (p = 0.26), and B (q = 0.14) and the genotypic frequencies were AA (p^2 = 0.08), AO (2pr = 0.31), BB (q2= 0.02), BO (2q r= 0.17), AB (2pq = 0.07), and OO (r^2 = 0.36) in Mecha. The samples collected from Yilmana Denssa had O (r=0.65), A (p=0.20) and B (q=0.15) allele frequencies and AA (p^2=0.04), AO (2pr=0.26), BB (q2=0.02), BO (2qr=0.20), AB (2pq=0.06) and OO (r^2=0.42) genotype frequencies. The cross total of the two sites had O (r=0.62), A (p=0.23) and B (q=0.15) of allele frequencies and the genotypic frequencies were AA (p^2=0.05), AO (2pr=0.29), BB (q^2=0.02), BO (2qr=0.18), AB (2pq=0.07) and OO (r^2=0.39) (Table 1).

Table 2: Estimates of fixation (f) and panmictic (p) indexes and observed vs. expected for abo gene in the study area (N=200).

Population ABO phenotypes	Obs. H (Ho)	Exp. H (He)	X^2	
O	0.39	0.39	0	
A	0.34	0.34	0	
B	0.21	0.20	0.02	
AB	0.06	0.07	0.07	
Rh(D)+VE	0.91	0.7	0	
Rh(d)-VE	0.09	0.3	0	
Population	Obs. H (Ho)	Exp. H (He)	F	P
Yilmata Densa total (ABO) N=100	0.55	0.67	17.10	83
Mecha Total N=100 (ABO)	0.52	0.67	22.43	78
Grand total N=200	0.54	0.67	20	80

P= manmictic index; F= fixation index X^2=chi-square, Obs. H (Ho)=observed hetrozygosity; Exp. H (He) = Expected hetrozygosity

Fixation indices (F) for ABO gene were 17 and 22 in the populations of the two sites. F for ABO gene was higher in Yilmana Denssa and Panmictic index (P) was the opposite of fixation index (F). Therefore, F plus P equals to unity. The P was 78 which were lower than Mecha site. The expected results ranged from 0.39 to 0.07 for ABO gene. On the other hand, the expected results were 0.91 and 0.09 for Rh negative and positive, respectively. The observed heterozygosity (Ho) for ABO genes ranged from 0.39 to 0.06. Ho for Rh negative was 0.7. From the results, AB blood group was significantly low. But there was no significant difference in the remaining blood groups at $\alpha = 0.05$. The observed heterozygosity (Ho) for ABO gene in the study area was 0.53 indicating large genetic variations in the populations (Table 2).

DISCUSSION

In this study, the frequencies were found to be in the order of i > IA > IB and IA =0.23, IB = 0.15 and i = 0.62 for ABO blood groups alleles and their genes, respectively. This is a common feature of Ethiopian population that is confirmed from previous studies. In the

study from Butajira, south Ethiopia, high allelic frequency of O blood group was reported which is in an agreement with our finding.[13] In a diverse ethnic background population of Ethiopian students, blood group study also showed similarity with our result.[12] Several studies indicate O blood group is most frequent in Africa which is very common in malaria endemic regions of the world.[15-17] Fixation indices (F) for ABO gene were 17 and 22 in the populations of the two (Mecha and Yilmana Densssa) sites. F for ABO gene was higher in Yilmana Densssa and Panmictic index (P) was lower than Mecha indicating low genetic variation for ABO gene. The overall F and P of the sites were 20 and 80 which is a characteristic feature of population with high genetic diversity since the ABO blood group distribution varies in different ethnic groups.[18] This is because Ethiopia is a country with more than 85 different ethnic groups having their own genetic diversity with culture of intermarriages among ethnic groups.[13] Expected frequency (genotypic frequency) was calculated from allele frequencies. The Hardy-Weinberg law states that both allele and genotype frequencies will remain constant from generation to generation in an infinitely large, interbreeding population in which mating is at random and there is no selection, migration and mutation. Under conditions of Hardy-Weinberg equilibrium, expected genotype frequencies may be derived from population allele frequencies. The observed heterozygosity (Ho) for ABO gene ranged from 0.39 to 0.07 indicating large genetic variations in these populations. Expected Hardy-Weinberg heterozygosity (He) was the same (0.6666) for all the populations as it was calculated assuming the occurrence of all the three alleles in equal frequency (q=1/k where k is the number of alleles).

In conclusion, blood group O dominated the study population followed by A, B and AB, respectively. The allele frequencies of A, B and O indicted O > A > B. This study confirmed that the ABO and Rh blood distributions were diverse in the study population. The population of Yilmana Densssa was highly inbred as well as more homogeneous as compared to the population of Mecha since the fixation index (F) of Mecha population was greater than that of Yilmana Denssa. The panmictic index (P) was greater in Mecha than that of Yilmana Densssa which revealed that population of Mecha were highly outbreeder with high heterozygosity (gene diversity) which confirms diverse nature of the populations of the study area. Therefore, the results of this study are of paramount importance for design and implementation of population genetics of the study area and can be a base for characterizing Ethiopian population genotypes. This study is limited both in its scope and area to phenotypic prediction of genotype and West Gojam, Northern Ethiopia part only.

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REFERENCES

1. Cavalli-Sforza L. Feldmann W. The application of molecular genetic approaches to the study of human evolution. Nat Genet. 2003;33:266-75.
2. Chakraborty S. Heterozygosity and Fixation Index for ABO Gene in Barak Valley populations. Not Sci Biol. 2011;3:7-11.
3. Reich D, Cargill M, Bolk S, Irel J, Sabeti P. Linkage disequilibrium in the human genome. Nature. 2001;411:199-204.
4. Tishkoff S. Kidd K. Implications of biogeography of human populations for 'race' and medicine. Nat Genet. 2004;36:21-7.
5. Jorde L, Wooding S. Genetic variation, classification and 'race'. Nat. Genet. 2004;36:28-33.
6. Yamamoto F. ABO blood group system ABH oligosaccharide antigens, anti-A and anti-B, A and B glycosyl transferases and ABO genes. Immuno hematol. 2004;20:3-17.
7. Choot E, Tax H. Rijnders J. Prenatal typing of Rh and Kell blood group system antigens: The edge of a watershed. Transfus Med. 2003;17:31-44.
8. Mourant E, Kopec C, Sobezak D. The distribution of the Human blood groups and other Polymorphisms. 1976; Oxford University Press, London.
9. Tekeste Z, Petros B. The ABO blood group and Plasmodium falciparum malaria in Awash, Metehara and Ziwayareas, Ethiopia. Malaria J. 2010;9:280.
10. Tadesse H. Tadesse K. Assessing the association of severe malaria infection and ABO blood groups in northwestern Ethiopia J Vector Borne Dis. 2013;50:292-6.
11. Oyenike A, Omolade O. Frequency distribution of ABO, RH blood groups and blood genotypes among the cell biology and genetics students of University of Lagos, Nigeria. African Journal of Biotechnology. 2006;5:2062-5.
12. Atire F. Distribution of ABO and Rh Blood Groups among Students of Some Ethnic Groups at Dilla University, Ethiopia. International Journal of Genetics and Genomics. 2015;3:8-19.
13. Tesfaye K, Petros Y, Andargie M. Frequency distribution of ABO and Rh (D) blood group alleles

in Silte Zone, Ethiopia, Egypt J. Med. Hum. Genet. 2014;16 (1).
14. Kaya H, Gündo du M, Akarsu E, Kiki I, Tekin B. The distribution of blood groups in Erzurum. Medical Journal of Atatürk University. 1999;31:20-2.
15. Bakare A, Azeez A, Agbolade O. Gene frequencies of ABO and rhesus blood groups and haemoglobin variants in Ogbomoso, South–West Nigeria. African Journal of Biotechnology. 2006;5:224-9.
16. Sadiq MA. Comparative study of some genetical triates of Ninawa population with Iraq population. J Education Scie. 1989;9:100-12.
17. Mouhaus A, Abbas H, Musa S, Mahawi K. A study of ABO blood group and Rhesus factor distribution among sample of Missan province population. Journal of Basrah Researches. 2010;36:48-53.
18. Chavhan A, Jadhao R, Pawar S. The study of Allelic Frequency of ABO and Rh D Blood Group among the Banjara Population of Akola District, Maharashtra, India. Journal of Anthropology. 2012;8:1973-2880.

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