

# Variations of ABO Blood Groups. Gene Frequencies in the Population of Sindh (Pakistan)

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The survey was carried out of allelic frequencies of ABO alleles in the population of different cities of Pakistan (Larkana, Jamshoro, Hyderabad, Karachi) and in different ethnic groups (Abro, Noohani and schedule cast Hindus). The analysis shows that there is no any constant pattern except the general fashion of blood group distribution i.e. the O group has highest frequency, A and B are in the mid and AB blood group is at least of all. Percentage distribution of ABO and their allelic frequencies are described in the districts/cities of the province of Sindh, Pakistan. Relationship between the distribution of ABO phenotypes and gene frequencies are discussed. A significant decrease in the AB phenotypes is seen in the cities. The average gene frequencies for ABO groups in the population of Sindh are  $0.1972 + 0.017$ ,  $0.2104 + 0.019$ ,  $0.5986 + 0.015$ , for A, B, O respectively. The average percentage distribution out of total 49061 cases the distribution of blood groups was A = 25.83%, B = 28.17%, O=37.78% and AB = 8.30%.

**Key words:** ABO blood groups, pattern of inheritance.

Blood transfusion used to be a risky operation as it is potentially dangerous drug. Some persons could accept the blood transfusion of other persons without complete injury, while with others, transfusion led to serious illness. Karl- Landsteiner found that people can be divided into four blood groups<sup>1</sup>, now called as groups A, B, O, and AB. Blood groups are inherited according to Mendel's law, through three variant forms of a gene (IA, IB, IO). Antigens are inherited as mendelian allelomorphs, A1, A2, and B being dominants<sup>2</sup>.

Brothers and sisters, parents and children may and quite often do, belongs to different blood groups. During world war 1, when many persons were in need of blood transfusions, it was discovered that the frequencies of four blood groups are rather different in persons native to different parts of the world. The incidence of individuals belonging to A,B,O and AB blood groups is different in different human populations. Boyd made an attempt to classify the racial groups of mankind according to incidence of known blood group<sup>3,4,5</sup>. Frequency distribution of blood groups has been investigated in different populations of the world and variations within population have been reported.

Several numerous investigations have been made and published since then specially in recent years to map the geographic distribution of blood groups. Shivaraman et al. (1971)<sup>6</sup> showed higher frequency for B allele in Punjab (India) than in Utter Pardesh (India). Regional and temporal variations in the ABO and Rh blood group allele frequencies in the country of Vesterbotten in northern Sweden were investigated by Beckman et al<sup>7</sup>. Imaizumi showed longitudinal and latitudinal clines for allele O in Great Britian. Shami and Kamboh investigated southwestward and northwestward clines for frequencies of alleles A,B,O and d in Punjab (Pakistan)<sup>8,9</sup>. Shami and Siddiqui investigated the clines for frequencies of alleles A,B,O and d in the Northern and Southern population of

Sindh (Pakistan)<sup>10</sup>. Turaeva et al discovered the geographic and ethnic differentiation of Turkmen population among five distinct and main Turkman tribes<sup>11</sup>.

The present study is also an attempt to establish the frequency distribution of ABO alleles in the population of different cities of Sindh (Pakistan). In different ethnic groups, the pattern of inheritance of blood group and the effect of different factors (genetic drift, migration breeding pattern) effecting the frequencies of ABO alleles. Walter et al investigated ethnic variability of ABO blood groups polymorphism in Iran and assumed that specific ABO allele frequencies found in different ethnic groups are connected with their different geographical origin<sup>12</sup>.

Proper understanding of human blood group system made possible the safer and beneficial transfusion. Nowadays, at present the research on blood groups system have different aims. Now it insighting the pattern of inheritance, frequencies of genes controlling blood groups, nature of the genes and their products, associations with different other characteristics and diseases. The mode of inheritance of blood group, the genes that control them and the frequencies of the genes in population is of great interest to geneticists. The frequency of different types of blood groups particularly the ABO is also important from anthropological point of view<sup>13</sup>. At present researches on blood group system is advancing in different directions. Comprehensive research work on the relationship between heredity blood factors and the disease have been done in the past. Different factors are found to be associated with skin psoriasis, within last few years it has been shown that there is an association between ABO groups and certain diseases<sup>14</sup>. Just a few years back this blood group system was considered to be natural as far as the fitness is concerned. The recent workers however, have shown that a definite relationship exist between the ABO system and a few

diseases<sup>15,16,17,18</sup>. Moreover the blood groups are not natural to selection and fitness. It is interesting to find out that this has been most clearly demonstrated in the case of disorders of upper intestinal tract where the secretion of blood group substances is the highest. The strongest relationship is between duodenal ulceration and group O<sup>19</sup>. Cancer of stomach in individuals with group 'A' than in groups 'O' and 'B'. There is also strong evidence of an association between pernicious anaemia and diabetes-mellitus with group 'A'.

Most individuals secrete water-soluble blood group substance in saliva. Blood typing helps in finding paternity. It has importance in medico-legal paternity cases<sup>2</sup>. Blood is most precious fluid in the body, a fact expressed in such common terms as "The life blood".

### Results

The mean for the percentage of ABO blood group out of 49061 of subjects belonging to different districts/cities of Sindh and different ethnic groups is A = 25.83%, B = 28.17%, O = 37.78% and AB = 8.30%. While for different cities, it is for Larkana A = 25.40, B = 32.09, O = 33.33 and AB = 9.18, for Karachi, A = 19.38, B = 19.62, O = 40.78 and AB = 20.21, for Jamshoro, A = 23.67, B = 24.65, O = 48.99 and AB = 2.69, for Hyderabad, A = 15.60, B = 21.79, O = 60.15 and AB = 2.99, and for different ethnic groups, e.g. for Abro A = 46.67, B = 40, O = 13.33 and AB = 0.0, for Noohani A = 31.25, B = 31.25, O = 18.75 and AB = 18.75, and for schedule casts (Hindu, Bheel and Coolhi) A = 18.86, B = 27.76, O = 49.11, and AB = 4.27.

The mean frequencies of ABO blood alleles and the chi-square contribution ( $\chi^2$ ) is A = 0.1972 + 0.017, B = 0.2104 + 0.019, O = 0.5986 + 0.015 and  $\chi^2$  = 108.0639 while for different cities is, for Larkana A = 0.1891 + 0.0030, B = 0.2315 + 0.0037, O = 0.5773 and  $\chi^2$  = 1.8065, for Karachi A = 0.1370 + 0.012, B = 0.1385 + 0.012, O = 0.6387 + 0.0155 and  $\chi^2$  = 598.5692 for Jamshoro A = 0.1525 + 0.0036, B = 0.1582 + 0.0027, O = 0.6999 + 0.0072 and  $\chi^2$  = 136.7518, for Hyderabad A = 0.0948 + 0.0032, B = 0.1294 + 0.0038, O = 0.7756 + 0.0040 and  $\chi^2$  = 0.1820 and for different ethnic groups is for Abro A = 0.4094 + 0.023, B = 0.3652 + 0.016, O = 0.3651 + 0.19 and  $\chi^2$  = 17.94, for Noohani A = 0.2741 + 0.0115, B = 0.2741 + 0.0115, O = 0.433 + 0.076 and chi-square = 1.1895 and for schedule cast (Hindu, Bheel and Coolhi), A = 0.1236 + , B = 0.176 + , O = 0.7008 + and  $\chi^2$  = 0.0083.

### Discussion

Since from the discovery of ABO blood groups by landsteiner the blood grouping remains important in past as well as at present it plays important multiple role not only in transfusion but in relation of transplant surgery, hereditary diseases, genetics and migration of different races of the world<sup>1</sup>. As the blood group ABO are inherited by three variant genes A, B and O of which the A and B are Co-dominant to each other and dominant over O. The

frequencies of these genes are variable in different nations, areas, and even in different ethnic groups of same area. According to Hardy-weinberg law the frequencies of the members of a pair of allele in a population are described by the expansion of the binomial equation. Mather, K, describes that if the members of the population are mating randomly and if no other factors are effecting allelic frequencies then the frequencies of various pairs of alleles should be very closed to those calculated with the Hardy-weinberg formula<sup>20</sup>. The factors that can effect the constancy of gene frequencies, are genetic drift (migration) and gene flow. The clines of gene frequency in man may be explained by the migration and inter-marriage of different races and also due to geographical differences in selection intensity with the diffusion effect of migration suggest that clines for ABO blood group gene frequencies in Japanese population show that the selection intensity may be primarily dependent on the temperature<sup>21,22,23</sup>. Shami and Kumboh<sup>5</sup> considered that the temperature differences in the northern and southern Punjab are quite appreciable and it may be that the selection intensity there depends on temperature. They also showed that the age of the subjects is not associated with the changes in frequencies of ABO genes and likewise sexes show no differences in the distribution of these blood group genes.

The present study is carried out to estimate the ABO blood groups percentages and frequencies of ABO blood allele in different cities of Sindh (Pakistan) and in some different ethnic groups.

The results show that collectively the blood group 'O' has got the highest percentage (37.78), blood group 'AB' has the lowest percentage (8.30). It confirms the already existing fact that in Pakistan 'B' and 'O' groups are more prevalent as compared to 'A' and AB<sup>24</sup>. The pattern is similar to different cities of Pakistan i.e. in Quetta and in Multan<sup>25,26</sup>. In Sindh, here the collective percentage of blood group 'O' is some what lower in comparison to other cities of Pakistan. The percentage of group 'O' in Larkana is less i.e. (33.33) and in the ethnic groups of Noohani (18.75), Abro (13.33) where the percentage of blood group 'O' is lower even than the blood group 'A' and 'B'. In similar manner the mean of ABO blood alleles has got similar pattern i.e. the 'O' allele has highest frequency (0.5986) followed by the 'B' allele (0.2104), slightly lower to it is of 'A' allele (0.1972). the pattern is similar to the distribution of these alleles in different cities of Pakistan i.e. Multan (O = 0.5469, B = 0.2692 and A = 0.1815)<sup>26</sup>. The pattern is also similar to the pattern in some different parts of India i.e. Muslim group of Gujrat (O = 0.61, B = 0.22 and A = 0.20), Paphia et al. (1981)<sup>27</sup>. Sikhs of Punjab (O = 0.61, B = 0.21 and A = 0.20) and Kangra Rajput's of Himachal Pardesh (O = 0.59, B = 0.21 and A = 0.20)<sup>28,29</sup>, while it is quite different from that in USA (O = 0.6708, B = 0.0708 and A = 0.2578), UK (O = 0.6856, B = 0.0560 and A = 0.2578) and Germany (O = 0.6557, B = 0.0792 and A = 0.2663) where the 'A' allele has significantly higher

frequency then 'B' allele but the blood 'O' allele has similarly highest frequency. In Vietnam a similar pattern to us, is found (O = 0.6708, B = 0.1894 and A = 0.1421). Though in general, collectively our total results follow the same pattern as of different cities of Pakistan and India but if we consider individually different cities and different ethnic groups we can see significant variations in the blood group percentages and in the frequencies of ABO blood alleles. The mean chi-square contribution ( $\chi^2=108.0639$ ) shows that overall our population is not at an equilibrium. The reason for that are may be mainly the genetic drift and gene flow. If we consider individually different cities the  $\chi^2$  values (Table 2) shows that city Larkana (1.8065) and Hyderabad (0.1820) are at an equilibrium state while the city of Karachi ( $\chi^2=598.5692$ ) and Jamshoro ( $\chi^2=136.7518$ ) has very

significant  $\chi^2$  values and are not at equilibrium state. This may be mainly because of the heterogeneity of the population of these cities. In the similar manner the ethnic groups of Noohani (1.1895) and Hindu (0.0083) are at equilibrium state while the ethnic group of Abro (17.9400) is not at equilibrium state, while all of these three have similar pattern of marriages i.e. they strictly marry in their own ethnic groups and equilibrium here can be reason at least for Hindus (Bheel and Coolhi) that the effect of gene flow (i.e. their mating habits) are neutralizing by the genetic drift (i.e. migration), they tend to live in the form of small tribes. In this way they may have developed a variable distribution from tribe to tribe which collectively gives them an equilibrium state. This pattern is also quite different to one those of Australian Aborigines who have only group 'O' and 'A'<sup>24</sup>.

Table:1 Distribution of Phenotypic Frequencies of ABO blood groups in the general population of Sindh (Pakistan).

City	Blood Group A	Blood group B	Blood group O	Bloodgroup AB
Larkana	25.40%	32.09%	33.33%	9.18%
Jamshoro	23.67%	24.65%	48.99%	2.69%
Hyderabad	15.60%	21.79%	60.15%	2.99%
Karachi	19.38%	19.62%	40.78%	20.21%
Abro	46.67%	40.0%	13.33%	0.0%
Noohani	31.25%	31.25%	18.75%	18.75%
Hindu(Bheel & Coolhi)	18.86%	27.76%	49.11%	4.27%
Mean	25.83%	28.17%	37.78	8.30%

Table:2 ABO blood allelic frequencies and Chi-square contributions in general population of Sindh (Pakistan).

City	A	B	O	X <sup>2</sup>	P
Larkana	0.1891+0.0030	0.2315+0.0037	0.5773+0.0082	1.8065	0.95
Jamshoro	0.1525+0.0036	0.1582+0.0027	0.6999+0.0072	136.7518	0.01
Hyderabad	0.0948+0.0032	0.1294+0.0038	0.7756+0.0040	0.1820	0.01
Karachi	0.1370+0.0120	0.1385+0.0120	0.6387+0.0155	598.5692	0.01
Abro	0.4094+0.023	0.3652+0.016	0.3651+0.19	17.9400	0.01
Noohani	0.2741+0.0115	0.2741+0.0115	0.4330+0.076	1.1895	0.99
Hindu	0.1236+0.017	0.1760+0.019	0.7008+0.015	0.0083186	0.95
Mean	0.1972+0.017	0.2104+0.019	0.5986+0.015	108.0639	0.01

(Table 3) shows the observed phenotypic frequencies, number and percentage observed with total number.

### Conclusion

The variability of the distribution of ABO blood group and their allelic frequencies, the presence of equilibrium, even in the presence of factor like genetic drift and gene flow shows that the ABO frequencies depends upon a complex interaction between the factors effecting the gene frequencies. It is not necessary that the presence of two factors will tighten the effects of each other but the presence of one can neutralize the effect of others. The variation in gene frequency may be due to random genetic drift or may be due to pattern of marriages<sup>30,31</sup>. The variation may be due to balanced effects of natural selection<sup>32</sup>. The possible explanation could be that selection is working against these phenotypes (The phenotypes having low frequencies for example AB)<sup>5</sup>. Mourant suggests that certain blood group phenotypes are

more susceptible to certain diseases resulting in loss of fertility<sup>33</sup>. This would be an important selective factor under certain conditions. Gene frequency will be found different from country to country, city to city, nation to nation and ethnic groups to ethnic groups, and even time to time. It also seems that the variability in the gene frequencies is inversely proportional to the size of population or number of persons in comparison.

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