

## VARIATIONS OF ABO AND Rh BLOOD GROUP ALLELE FREQUENCIES IN THE POPULATION OF SIND (PAKISTAN)

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**Abstract.** - Percentage distribution and allele frequencies of ABO and Rh-phenotypes are described in the cities of the Sind province, Pakistan. Sex differences and relationship between the distribution of ABO and Rh-phenotypes are discussed. A significant differences in the distribution of ABO-phenotypes in the males and females are seen in Jacobabad, Mirpur Khas, Nawabshah, Sukkur and Thatta. Average allele frequencies for ABO and Rh groups in the population of Sind are A=0.17; B=0.22; O=0.61; d=0.15.

**Key words:** Blood group, allele frequency.

### INTRODUCTON

**T**he genotype and phenotype frequencies for ABO and Rh blood groups have been investigated by different authors from various parts of the world and found that the incidence of blood groups varies from one race to another. In European population the frequency of allele A is higher than the frequency of allele B as described in Swedish (Beckman, 1972), Lipari-Italian (Warwick *et al.*, 1972) and Byellorussian and Poles (Oreknova, 1975) populations. Same is reported in Antham (Chileotin) Indians (Alfered *et al.*, 1970). In the populations of north and west Africa the frequency of allele A is still higher than allele B as reported for Aswan and Nubians (Azim *et al.*, 1974), North sinai (Hamza *et al.*, 1976) and Nigerian (Odeigah, 1990) populations. In contrast in southeast Asian populations the frequency of allele B is noticeably greater than allele A as reported in Rajasthan (Hurkat *et al.*, 1971) and Punjab, Pakistan (Shami and Kamboh, 1979). The variation in ABO distribution envisages not only the frequency distribution of phenotypes and genotypes, but also studies were carried out to unveil other variables in relation to these blood groups. Such studies include differential fertility and mortality in relation to ABO groups (Bennt and Walker, 1955), effects of natural selection on ABO locus (Chung and Morton, 1961), ABO-incompatibility (Lauriksen *et al.*, 1975), association with goiter (Harison *et al.*, 1976), thyroid cancer (Hernandez *et al.*, 1980), Breast cancer (Levine *et al.*, 1981), Cardiac infection (Pdatt *et al.*, 1985), carcinoma of stomach (Mecklin *et al.*, 1988), lung cancer (Cerny *et al.*, 1993), Linkage between ABO and nail patella (Renwick and Lawler, 1955) and Rh and marfan syndrome (Mace and Margaret, 1979).

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The present investigation shows the distribution of phenotypes and genotypes of ABO and Rh blood groups in the population of Sind (Pakistan) based on data from 13 cities.

### MATERIALS AND METHODS

The data are based on records of Blood Transfusion Centers and Hospitals in different cities of Sind (Pakistan). The data were collected from Badin, Dadu, Hyderabad, Jacobabad, Karachi, Khairpur, Larkana, Mirpur Khas, Nawabshah, Sanghar, Shikarpur, Sukkur and Thatta ( Fig. 1 ). The individuals tested for ABO system were also typed for Rh group i.e., D-positive and D-negative. Chi-square tests were carried out for different analysis.

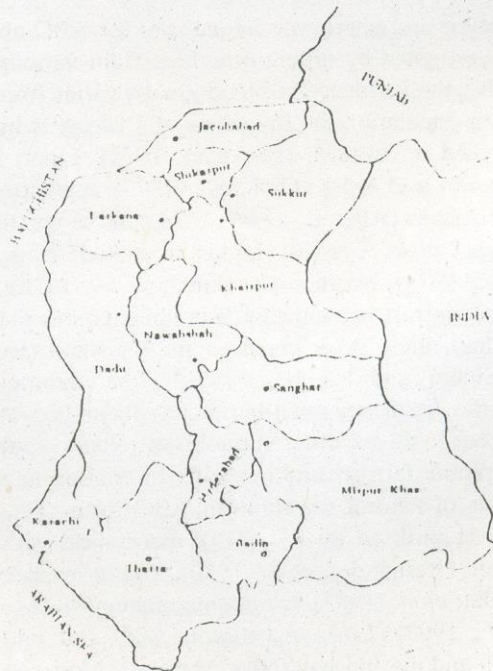


Fig. 1. Map of Sind (Pakistan) showing the cities of data collection.



## BLOOD GROUP ALLELE FREQUENCIES

## RESULTS

The highest percentage for A-phenotype is seen in Shikarpur (28.68) followed by Thatta (27.84), Nawabshah (27.64), Mirpur Khas (27.56), Jacobabad (26.57) and Sanghar (26.43). A remarkably low percentage for A-phenotype is seen in Dadu (22.10), Sukkur (21.97) and Larkana (18.62). Group B-phenotype shows highest percentage in Karachi (36.04) followed by Sukkur (35.59), Hyderabad (34.86) and Jacobabad (34.77). The lowest percentage values are seen in Dadu (28.15) and Shikarpur (23.23).

The highest percentage for AB-phenotype is observed in Karachi (9.36) followed by Mirpur Khas (8.64), Sanghar (8.41), Hyderabad (7.92) and Sukkur (7.62). The lowest percentage values are seen in Jacobabad (4.50), Thatta (4.10) and Larkana (3.90).

The O-phenotype shows higher percentage compared with A, B and AB-phenotypes. The highest percentage of O-phenotype is observed in Larkana (47.65) followed by Dadu (43.49) and Shikarpur (43.09). The lowest values are seen in Mirpur Khas (31.92) and Karachi (30.65).

The percentage distribution of ABO phenotypes in males and females is almost the same in different cities of Sind. Appreciable differences in A-phenotypes are seen in Mirpur Khas ( $\sigma\sigma$  30.69;  $\text{♀♀}$  21.96), Thatta ( $\sigma\sigma$  26.56,  $\text{♀♀}$  33.04) and Khairpur ( $\sigma\sigma$  27.64;  $\text{♀♀}$  21.75). In B-phenotype differences are observed in Jacobabad ( $\sigma\sigma$  29.33;  $\text{♀♀}$  40.37) and Mirpur Khas ( $\sigma\sigma$  29.27;  $\text{♀♀}$  37.06). AB-phenotypes show difference in Mirpur Khas ( $\sigma\sigma$  5.83;  $\text{♀♀}$  13.14), Badin ( $\sigma\sigma$  5.40;  $\text{♀♀}$  8.97) and Sukkur ( $\sigma\sigma$  6.37;  $\text{♀♀}$  9.43). In O-phenotype a difference ranging between 3.00-8.00 can be seen in most of the cities except prominent differences in Thatta ( $\sigma\sigma$  37.31;  $\text{♀♀}$  26.25) and Shikarpur ( $\sigma\sigma$  47.17;  $\text{♀♀}$  37.67).

Chi-square tests were carried out to find differences in the distribution of male and female ABO-phenotypes. Significant differences are observed in Jacobabad ( $X^2_3=15.35$ ;  $p<0.01$ ), Mirpur Khas ( $X^2_3=40.01$ ;  $p<0.001$ ), Nawabshah ( $X^2_3=15.15$ ;  $p<0.01$ ), Sukkur ( $X^2_3=10.24$ ;  $p<0.02$ ) and Thatta ( $X^2_3=11.83$ ;  $p<0.01$ ).

Chi-square tests were carried out to find goodness of fit to Hardy-Weinberg expectation in relation to ABO-phenotypes (Table I). The results are statistically non-significant showing agreement with Hardy-Weinberg expectation. Highly significant deviation from Hardy-Weinberg expectation is seen in Jacobabad ( $X^2_1=21.08$ ;  $p<0.001$ ), Nawabshah ( $X^2_1=14.73$ ;  $p<0.001$ ) and Thatta ( $X^2_1=26.24$ ;  $P<0.001$ ).

Data were also arranged to see distribution of ABO-phenotypes in combination with Rh-group. The different combination in which data were arranged are Rh<sup>+</sup>/A, Rh<sup>+</sup>/B, Rh<sup>+</sup>/AB, Rh<sup>+</sup>/O and Rh<sup>-</sup>/A, Rh<sup>-</sup>/B, Rh<sup>-</sup>/AB, Rh<sup>-</sup>/O.



**Table I.** Allele frequencies and goodness of fit to Hardy-Weinberg expectation of ABO phenotypes in the cities of Sind (Pakistan)

Locality	Blood Groups				Allele frequencies			X <sup>2</sup> <sub>1</sub>	P
	A	B	AB	O	A	B	O		
Badin	155	209	46	235	0.17	0.22	0.61	0.36	n.s
Dadu	307	391	87	604	0.15	0.19	0.66	0.92	n.s
Hyderabad	738	1070	243	1018	0.18	0.24	0.58	3.52	n.s
Jacobabad	266	348	45	342	0.17	0.22	0.61	21.08	<0.001
Karachi	1054	1586	412	1349	0.18	0.26	0.56	1.30	n.s
Khairpur	209	268	54	325	0.17	0.21	0.62	1.16	n.s
Larkana	234	375	49	599	0.12	0.19	0.69	1.47	n.s
Mirpur Khas	391	455	120	453	0.20	0.23	0.57	1.43	n.s
Nawabshah	681	795	162	826	0.19	0.22	0.59	14.73	<0.001
Sanghar	355	397	113	478	0.19	0.21	0.60	0.38	n.s
Shikarpur	195	149	34	293	0.19	0.15	0.66	1.05	n.s
Sukkur	331	537	115	526	0.16	0.25	0.59	0.46	n.s
Thatta	326	385	48	412	0.18	0.21	0.61	26.24	<0.001
<b>Average</b>					<b>0.17</b>	<b>0.22</b>	<b>0.61</b>		

n.s. = Non-significant

A higher percentage of Rh<sup>+</sup>/A-phenotype is seen in Shikarpur (28.80), Nawabshah (27.90) and Thatta (27.91). A higher percentage of Rh<sup>+</sup>/B-phenotype is seen in Karachi (36.26), Sukkur (35.56) and Hyderabad (35.07). Rh<sup>+</sup>/AB-phenotype shows higher percentage in Karachi (9.31), Mirpur Khas (8.58), Sanghar (8.46) and Hyderabad (8.23). Rh<sup>+</sup>/O-phenotype shows higher percentage in Larkana (47.74). Rh<sup>-</sup>/A-phenotype shows higher percentage in Mirpur Khas (32.89) and Sukkur (27.54). A higher percentage of Rh<sup>-</sup>/B-phenotype is seen in Badin (66.67) and Dadu (50.00). Rh<sup>-</sup>/AB-phenotype shows higher percentage in Khairpur (100.00), Sanghar (71.42) and Shikarpur (66.67).

A higher percentage of Rh<sup>+</sup>/ABO-phenotype is seen in males as compared to females in all cities except in Dadu (64.17), Khairpur (54.80) and Hyderabad (63.21) where females have higher percentage when all phenotypes are taken together. Rh<sup>+</sup>/B (33.98) and Rh<sup>+</sup>/AB (9.06) females from Badin, Rh<sup>+</sup>/A (23.15) and Rh<sup>+</sup>/AB (6.74) females from Dadu, Rh<sup>+</sup>/AB females (8.62) from Hyderabad, Rh<sup>+</sup>/B (40.33) females from Jacobabad, Rh<sup>+</sup>/A (24.71) and Rh<sup>+</sup>/B (10.42) females from Karachi, Rh<sup>+</sup>/B (33.55) and Rh<sup>+</sup>/AB (7.27) females from Khairpur, Rh<sup>+</sup>/A (22.03), Rh<sup>+</sup>/B (30.39) and Rh<sup>+</sup>/AB (5.73) females from Larkana, Rh<sup>+</sup>/B (37.58) and Rh<sup>+</sup>/AB (12.53) females from Mirpur Khas, Rh<sup>+</sup>/A (29.90) and Rh<sup>+</sup>/AB (7.08) females from Nawabshah, Rh<sup>+</sup>/A (31.21) and Rh<sup>+</sup>/AB (10.83) females from Sanghar, Rh<sup>+</sup>/A (24.14) and Rh<sup>+</sup>/AB (9.25) females from Sukkur, Rh<sup>+</sup>/A (33.19), Rh<sup>+</sup>/B (5.67) females from Thatta show higher percentage for respective phenotypic combination compared with that of males.



## BLOOD GROUP ALLELE FREQUENCIES

Chi-square tests were carried out to see relationship of ABO-phenotypes in combination with Rh-positive and Rh-negative blood groups. Statistically significant results are obtained from Mirpur Khas ( $X^2_3=11.08$ ;  $p<0.02$ ), Nawabshah ( $X^2_3=10.83$ ;  $p<0.02$ ) and Thatta ( $X^2_3=24.02$ ;  $p<0.001$ ).

Chi-square tests were carried out to compare the distribution of ABO-positive phenotypes ( $A^+$ ,  $B^+$ ,  $AB^+$ ,  $O^+$ ) and ABO-negative phenotypes ( $A^-$ ,  $B^-$ ,  $AB^-$ ,  $O^-$ ) in males and females. Statistically significant results for ABO-positive phenotypes are observed in Jacobabad ( $X^2_3=15.58$ ;  $p<0.01$ ), Mirpur Khas ( $X^2_3=25.77$ ;  $p<0.01$ ), Nawabshah ( $X^2_3=9.02$ ;  $p<0.05$ ), Sukkur ( $X^2_3=9.75$ ;  $p<0.02$ ) and Thatta ( $X^2_3=11.01$ ;  $p<0.02$ ). Only two statistically significant results, regarding ABO-negative phenotypes, are obtained from Mirpur Khas ( $X^2_3=22.36$ ;  $p<0.001$ ) and Nawabshah ( $X^2_3=14.28$ ;  $p<0.01$ ).

Table II shows the allele frequencies for allele d in males and females, sexes combined in different cities of Sind. There are regional differences in the distribution of allele d. The highest frequency of allele d is seen in Mirpur Khas (0.32) and lowest in Thatta (0.03). In males and females the highest frequency for allele d is observed in Mirpur Khas (0.33 and 0.30 respectively). No case of Rh-negative males and females is reported from Thatta and Shikarpur respectively.

**Table II. Frequencies of allele d in males, females and sexes combined in the cities of Sind (Pakistan).**

Locality	Males	Females	Sexes combined
Badin	0.13	0.10	0.12
Dadu	0.04	0.03	0.04
Hyderabad	0.25	0.28	0.27
Jacobabad	0.11	0.12	0.11
Karachi	0.24	0.23	0.24
Khairpur	0.05	0.05	0.05
Larkana	0.12	0.11	0.12
Mirpur Khas	0.33	0.30	0.32
Nawabshah	0.31	0.22	0.27
Sanghar	0.06	0.11	0.07
Shikarpur	0.09	0.00	0.07
Sukkur	0.21	0.22	0.21
Thatta	0.00	0.07	0.03
<b>Average</b>	<b>0.15</b>	<b>0.14</b>	<b>0.15</b>



### DISCUSSION

The present investigation shows differences in allele frequencies in different parts of the Sind (Pakistan). Lowest frequency of allele A is seen in Larkana and highest in Mirpur Khas. Allele B showed lowest frequency in Shikarpur and highest in Karachi. The lowest frequency of allele O is observed in Karachi and highest in Larkana. The variations in gene frequency may be due to random genetic drift (Wright, 1931; 1940 and Turaeva *et al.*, 1984). The variations in gene frequency may be maintained due to balanced effects on natural selection (Ford, 1945). Thompson (1972) investigated that changes in population structure can produce large effect on gene frequencies in one generation as any normal selective force. At the moment it is not possible to show which of the three forces is more effective in the present population. Considering the marriage types, most of the marriages are between the relatives than between persons not known to be related. This would obviously encourage the incidence of certain alleles, but at the same time the respective phenotypes may show susceptibility to a certain disease which would have a selective effect on phenotypes.

The ABO-phenotypes show complete agreement to Hardy-Weinberg expectation except in Jacobabad, Nawabshah and Thatta (Table I). Shami and Kamboh (1979) showed highly significant deviation from Hardy-Weinberg expectation in four cities of the Punjab, Pakistan (Gujranwala, Multan, Sahiwal and Sialkot). The data show that the main cause of significant deviation from Hardy-Weinberg expectation is AB individuals, showing significant differences between observed and expected phenotypic values. This could be due to misclassification, inbreeding etc., (Beckman *et al.*, 1972). The probability of significant deviation from Hardy-Weinberg expectation due to misclassification seems to be low since discrepancy is seen only in AB individuals at three places. The possible explanation could be that selection is working against these phenotypes. Mourant (1974) suggest that certain blood group phenotypes are more susceptible to certain diseases resulting in loss of fertility. This would an important selective factor under certain conditions.

The data lack information regarding the association to certain disease with blood group, however, evidence derived from the existing data show that AB phenotype are more susceptible to smallpox (Vogal & Chakravarti, 1966), tuberculosis (Jain, 1970), chicken pox (Hepp *et al.*, 1975), cardiac infection (Pdatt *et al.*, 1985) and lung cancer (Cerny *et al.*, 1993).

The distribution of ABO-phenotypes in the two sexes show significant differences in five cities. Shami and Kamboh (1979) showed significant differences in the distribution of ABO-phenotypes in two sexes in the six cities of the Punjab (Pakistan).

The lowest frequency for allele d is seen in Thatta (0.03) and the highest in Mirpur Khas (0.32). The data do not show any record of Rh-negative male from Thatta and female.



## BLOOD GROUP ALLELE FREQUENCIES

from Shikarpur. Singh *et al.* (1974) showed absence of Rh-negative in Tibetans. No case of Rh-negative was recorded from Muzafargarh (Shami and Kamboh, 1979).

The population of Assam shows similarity with regard to ABO blood group frequencies with Karachi (Gupta and Sarthak, 1979). The populations are lying at the border of two countries. Although there is no mixing between two populations since the establishment of Pakistan, but still the two populations are maintaining more or less the same distribution of ABO blood group alleles. Shami and Kamboh (1979) showed resemblance of frequencies of alleles B and O in the population of Rajasthan (India) and Bahawalpur (Pakistan).

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