

# Genetic Variation of Blood Group Polymorphism among an Endogamous Human Population from Andhra Pradesh, India

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## Abstract

Genetic polymorphic traits like blood groups are widely used genetic markers in human population genetics. The present paper reports the genetic variation in phenotype and allele frequencies of ABO and Rh (D) blood groups among Kshatriyas, an endogamous population from Visakhapatnam district, Andhra Pradesh, India. Blood samples of 100 unrelated Kshatriyas, individuals were screened for ABO and Rh (D) blood groups. The order of occurrence of ABO phenotypes is  $O > B > A > AB$ . The corresponding allele frequencies of O, A and B are 0.420, 0.155 and 0.415, respectively. The allele frequency of  $D$  (0.990) is more than  $d$  (0.010). The present results were compared with the other populations to understand the population variations.

**Keywords:** Genetic polymorphism, Genetic variation, Blood groups

## Introduction:

ABO and Rhesus (Rh) blood group systems are hereditary characters and are used widely in human population genetic studies. The knowledge of the distribution of ABO and Rh blood groups at local and regional levels are helpful in the effective management of blood banks and in blood transfusion services. The use of blood group systems are conventionally referred, to as inherited antigens detected on the red cell surface by specific antibodies. Because of this individuality and because of the rigid manner of their inheritance in different populations, blood groups can be applied to various issues such as identifying disputed parentage and paternity.<sup>1</sup> Further, blood group systems are useful to determine the genetic variation within and between the populations. In human species, the best-known example of multiple allelism is the blood groups. The availability of very sensitive immunological tests has led to recognizing numerous alleles at a

particular locus. In some groups of multiple alleles, certain heterozygotes show dominance or intermediary and other heterozygotes with alleles and with qualitatively different effects show both effects, simultaneously. Thus in blood groups, the combination of dominance and codominance are found.<sup>2</sup> The latest literature about the blood group markers revealed that the blood groups systems are inherently polymorphic and show variability across the populations.<sup>3-7</sup> During the last four decades, numerous studies have been carried out on the genetic composition of various endogamous population groups in India.<sup>8,9</sup> However, genetic studies among Kshatriyas are very limited. Hence, in the present study, the distribution of ABO and Rh blood groups has been reported among the Kshatriyas, a forward caste of Visakhapatnam district, Andhra Pradesh, India.

Kshatriyas is one of the most predominant and largest Telugu speaking castes in Andhra Pradesh.

Traditionally, it is a worrier community in Hindu society, and hierarchically, they are superior to many other castes, except Brahmins in Andhra Pradesh. They are treated as higher caste group in coastal Andhra Pradesh. Kshatriyas are forward caste in line of social caste ranking. Rajulu, suryavamsa rajulu and chandravamsa rajulu are the main synonymous to Kshatriyas. Kshatriyas selected for the present study is found to be distributed mainly in the Visakhapatnam district of Coastal Andhra Pradesh. At present Kshatriyas are accustomed to agriculture, businesses, and also switched over to government jobs and also settled in trade and industry is their main stay of economy. Monogamy is the general pattern of marriage. Literacy rate is low compared to the neighboring caste populations. Joint families are more common. They speak Telugu, a Dravidian language. The ethnographic profile of Kshatriyas is available elsewhere.<sup>10</sup>

### Materials and Methods:

The blood samples were drawn from randomly sampled 100 healthy unrelated men and women of Kshatriyas, population of Visakhapatnam district in Coastal Andhra Pradesh. The blood samples were collected according to the methods of Race and Sanger.<sup>1</sup> The 2 to 3 drops of direct blood from the finger tip were taken into the sterile test tubes containing 0.9% saline solution. RBCs are washed with normal saline solution and 5% red cell suspension was prepared. Following the standard techniques with adequate controls, the RBCs were grouped for ABO blood groups and Rh types, through agglutination method [11]. The antisera used were anti-A, anti-B and anti-D antisera supplied by Ethnor Limited, Bombay, India.

Gene (allele) frequencies are calculated by following Bernstein's formula,<sup>12</sup> explained along with corrections by Mourant et al.<sup>11</sup> All these statistics were done as illustrated by Balakrishnan,<sup>13</sup> and these statistics are presented briefly hereunder. Phenotype frequencies of blood group systems are presented as simple percentages. The gene frequencies of ABO blood groups were calculated as:

$$p=1-\sqrt{0+B}; q=1-\sqrt{0+A} \text{ and } r=\sqrt{0}$$

A, B and 0 denote the proportion of the population in various groups; p is the frequency of gene A, q is the frequency of gene B and r is the frequency of the gene O. If the total of p + q + r are not adding up to one, the deviation from one is noted as D, an improved estimate of the gene frequencies given by Bernstein is

$$\begin{aligned} p' &= p(1 + \frac{1}{2}D) \\ q' &= q(1 + \frac{1}{2}D) \\ r' &= (r + \frac{1}{2}D)(1 + \frac{1}{2}D) \end{aligned}$$

The allele frequencies of Rh blood group were estimated as following:

Gene frequency of d =  $\sqrt{d}$ , where d = proportion of the population in Rh d group; gene frequency of D = 1-d.

### Results and Discussion:

ABO and Rh genes and phenotypes vary widely across races and geographical boundaries despite the fact that the antigens involved are stable throughout life. The resultant polymorphism remains important in population genetic studies, estimating the availability of compatible blood. The present study is, therefore, useful in providing information on the status of ABO and Rh blood group distribution among the Kshatriyas. The distribution of ABO blood groups is presented in Table 1. The group 'O' (42%) is the most frequently encountered phenotype in the population under the study. This observation is in accordance with the previous reports from other parts of Andhra Pradesh. With regard to the other phenotypes of ABO blood groups, the frequency of group 'B' is 40 %, and group 'A' is 13%, while 'AB' group was the least occurred phenotype with a frequency of 5%. The order of ABO percentage frequencies is O > B > A > AB. With regard to distribution of allele frequencies among Kshatriyas, allele O is observed with the highest frequency (0.420) than B (0.415) and B (0.155). Reviewing the ABO phenotype frequencies of other caste populations of Andhra Pradesh, it can be observed that a majority of populations recorded higher frequencies of blood group O than either B or A,

while a few reported higher frequencies of B.<sup>14</sup> Table 2 shows the frequency of Rh (D) phenotypes among the study population. In the present study, the frequency of Rh-D positive phenotype is 99% and 1% were Rh negative and the allele frequencies of *D* and *d* are 0.99 and 0.01, respectively. This is in general with other Andhra caste populations who reported low frequency of Rh (D) negative gene,<sup>15</sup> and several populations of Andhra Pradesh reported the absence of this gene.<sup>16</sup>

**Table 1: Distribution of the ABO blood group and their allele frequencies among Kshatriya caste, Andhra Pradesh, India:**

<i>Phenotype</i>	<i>Phenotypic frequency</i>	<i>Allele Frequency</i>
O	42.0	0.420
A	13.0	0.155
B	40.0	0.415
AB	5.0	

**Table 2: Distribution of the Rh (D) blood group and their allele frequencies among Kshatriya caste, Andhra Pradesh, India**

<i>Phenotype</i>	<i>Phenotypic frequency</i>	<i>Allele Frequency</i>
Rh (D) + ve	99.0	0.990
Rh (D) – ve	1.0	0.010

In the present study, the frequency of O blood group is more than other groups. The O blood type (due to absence of both A and B alleles) is common among many populations of the world. It is particularly high in frequency among the indigenous populations of Central and South America, where it very nearer to 100%. It also is relatively high among Australian Aborigines and in Western European population of Celtic origin. Some anthropologists postulated that the allele A evolved earliest, followed by allele O, by the deletion of a single nucleotide, and then B. This chronology resulted the presence of people

worldwide with each blood type. It is consistent with the accepted patterns of early population movements and varying prevalent blood types in different parts of the world: for instance, B is very common in populations of Asian descent, but rare in populations of Western European descent. Another theory states that there are four main lineages of the ABO gene and that mutations creating type O have occurred at least three times in humans.<sup>17</sup> The continued presence of the O alleles is hypothesized to be the result of balancing selection.<sup>17</sup>

### Conclusion:

An earlier study on genetic distance analysis of Andhra castes based on ABO and Rh blood group genotypes revealed a pattern, discernable in clustering of castes of same hierarchy like forward castes forming close clusters with other forward castes, and backward castes again forming close clusters with other backward castes.<sup>14,18</sup> It is to be mentioned that each caste population is endogamous from times immemorial which automatically rules out genetic admixture with other populations with a few possible exceptions. The initial differentiation of these communities is primarily based on occupations. So to start with, many of the present day caste populations might have originated from a set of common ancestors, but this differentiation might have taken place long ago, after which, there was no significant genetic admixture.

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