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Y Chromosome Polymorphism Among Five Different Ethnic Groups of Haryana

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ABSTRACT Size variability of the Y chromosome among five different ethnic groups of Haryana, *viz*. Jat, Ahir, Saini, Kamboj and Ror, was evaluated during the present investigation. As many as four Y indices were calculated using four different sets of chromosomes as "Standard chromosomes" namely 13-15 (Y/D index), 16-18 (Y/E index), 19-20 (Y/F index) and 21-22 (Y/G index) and compared with different populations of the world. Long Y chromosome was recorded in one individual each of the Saini and Kamboj samples. Its frequency was noted to be 5% in each of these groups. The "t-test" was applied to find out whether inter group differences are significant or no-significant. Various aspects of the Y chromosome polymorphism have been disucssed in the light of existing data.

INTRODUCTION

Cytogenetic studies on the general population were initiated by Jacob et al. (1961) and are desirable for a number of reasons. Cytogenetic investigations help to determine the frequency of variations in the morphology of chromosomes (Saski et al., 1963; Chandra and Hungerford, 1963; Chapelle et al., 1963 a, b). Polymorphic variants not only characterize whole chromosomes but also part of the chromosome.

Chromosomal variants can be used as a marker in population studies as well as for other studies (Muller et al., 1972). Different investigators have asessed the Y chromosome differently from other acrocentric chromosomes on morphological grounds (Chu and Giles, 1959; Tjio and Puck, 1958; Patau, 1960). A large Y chromosome seems to be a common feature in normal populations without causing developmental disorders. However, in spite of the importance and significance of cytogenetic investigations not a single ethnic group from Haryana population has been subjected to such study todate. To fill up these lacunae in our knowledge the present investigations were undertaken, to study the morphological variation in the length of Y chromosome among five different ethnic groups, *viz*. the Jat, Ahir, Saini, Kamboj and Ror.

MATERIALS AND METHODS

Peripheral blood cultures of 20 normal individuals belonging to five different ethnic groups of Haryana were set up to study Y-chromosome polymorphism. Photomicrographs of ten well spread metaphases were used for preparing the karyotypes of each individual. Length of the chromosomes was measured using a dial calliper (Mitutuyo Japan). The data obtained for the chromosomal aberrations and percentage relative length were evaluated using various statistical tests (Colton, 1980).

In studying Y chromosome polymorphism, particularly the length variation of the Y chromosome four Y indices were calculated using four different sets of chromosomes as "standard chromosomes" namely D, E, F and G. The "Y indices" were calculated as follows:

$$\text{Y index} = \frac{\text{Total length of Y chromosome}}{\text{Average total length of "Standard Chromosome."}}$$

Long Y chromosomes were noted when their length exceeded that of the chromosome 19.

RESULTS

The mean, standard deviation, coefficient of variation and variance of four different indices were calculated and are shown in table 1.

The mean Y/D index value was found to be 0.445±0.1200 among the Jat; 0.4800±0.1467 among the Ahir; 0.5340±0.1524 among the Saini; 0.5130±0.2068 among the Kamboj and 0.4835±0.1921 among the Ror.

The mean Y/E index value was found to be 0.5045±0.1565 among the Jat; 0.5180±0.1377 among the Ahir; 0.5700±0.11421

Table 1 : Mean, standard deviation coefficient of variance and variance of the four Y indices Y/D, Y/E, Y/F and Y/G of five different ethnic groups together with different populations

Index	*Group*	*No.*	$\overline{X}$	*S.D.*	*C.V.*	*Variance*
Y/D	Jat	20	0.4445	0.1200	17.0	0.01440
	Ahir	20	0.4800	0.1467	20.5	0.02155
	Saini	20	0.5340	0.1524	18.5	0.02329
	Kamboj	20	0.5130	0.2069	20.3	0.04283
	Ror	20	0.4835	0.1921	19.7	0.03964
	Punjabis	100	0.5984	0.0712	11.9	0.00506
	Rajputs	100	0.6504	0.0831	12.8	0.00690
	Japanese	11	0.5920	0.0750	12.7	0.00562
Y/E	Jat	20	0.5045	0.1565	18.0	0.02465
	Ahir	20	0.5180	0.1377	16.5	0.01903
	Saini	20	0.5700	0.1421	17.9	0.02026
	Kamboj	20	0.5385	0.1868	17.6	0.03494
	Ror	20	0.5485	0.1903	13.6	0.03628
	Punjabis	100	0.6765	0.0812	12.0	0.00659
	Rajput	100	0.7479	0.1214	16.2	0.01473
	Japanese	11	0.7050	0.0580	8.2	0.00336
	Finnish	30	0.7306	0.0658	9.0	0.00432
Y/F	Jat	20	0.6215	0.1620	20.0	0.02630
	Ahir	20	0.6710	0.1669	14.8	0.02789
	Saini	20	0.7080	0.1793	15.3	0.03223
	Kamboj	20	0.6370	0.1976	13.0	0.03917
	Ror	20	0.6495	0.1846	12.4	0.03425
	Punjabis	100	0.7971	0.1025	12.9	0.01050
	Rajput	100	0.9019	0.1429	15.8	0.02042
	Indians	20	0.8842	0.1081	12.2	0.1168
	Japanese	20	1.0019	0.1527	15.2	0.02331
	Negroes	20	0.9223	0.0868	9.4	0.00753
	Jews	20	0.9416	0.0998	10.6	0.00996
	Non-Jews	20	0.8621	0.0887	10.3	0.00786
	Germans	40	0.9856	0.0828	8.4	0.00684
	Japanese	11	0.8730	0.0510	5.8	0.00260
Y/G	Jat	20	0.9120	0.2509	16.4	0.05804
	Ahir	20	0.9490	0.2066	19.7	0.04277
	Saini	20	0.9835	0.2206	20.4	0.04879
	Kamboj	20	0.9495	0.2751	18.9	0.06574
	Ror	20	0.9265	0.2185	12.5	0.04770
	Punjabis	100	1.0272	0.1013	9.8	0.01026
	Rajput	100	1.1597	0.1718	14.8	0.02951
	Japanese	11	1.1130	0.0570	5.1	0.00324

among the Saini; 0.55385±0.1868 among the Kamboj and 0.5458 ± 0.1903 among the Ror.

The mean Y/F index value was found to be 0.6215±0.1620 among the Jat; 0.6710± 0.1669 among the Ahir; 0.7080±1793 among the Saini; 0.6370±0.1976 among the kambonj and 0.6495±0.1846 among the Ror.

The mean Y/G index value was found to be 0.1920±0.2409 among the Jat; 0.9490 ± 0.2066 among the Ahir; 0.9835±0.2206 among the Saini; 0.9495±0.2751 among the Kamboj and 0.9265±0.2185 among the Ror.

The values of Y/D, Y/E, Y/F and Y/G indices, were found to be the highest among the Saini and the minimum among the Jat.

DISCUSSION

Morphological variability of the human chromosome is very well known. It occurs in about 5% of the general population (Ferguson-Smith, 1972). This variability is especially evident in the distal part of the long arm of the Y chromosome (Lubs and Ruddle, 1971). The Y chromosome in man is mostly heteropycnotic and carries in its longer arm a large achromatic region (Sasaki and Makino, 1963). The heteropycnosis is coupled with the late replication of DNA (German, 1962, 1964; Schmid, 1963; Kikuchi and Sandberg, 1965). These phenomena are considered to be the manifestation of a genetically inert region (Klevecz and Hsu, 1964). Becuase of its genetically inert nature, size variability of the Y chromosome is possible, since structural alterations in such inert part of the Y are not reflected in phenotypical modifications. Moreover, the inert chromosomes are subject to loss or variation in both form and size.

Variations in the length of the Y chromosome were found to be relatively common in the measurements reported at the Denver Conference (1960). The Y chromosome varies more in length from subject to subject than other chromosomes (Court Brown et al., 1966; Walzer et al., 1969). Numerous reports of individuals and populations with variant Y chromosome have appeared since the Denver conference. Beginning with the finding of 15% increase in the length of the Y chromosome in a highly intelligent man (Bender and Gooch, 1961), a considerable number of cases are available in the literature with unusually long Y chromosome. Jacob and Harnden (1961) observed long Y in a Down's syndrome. Bishop et al. (1962) observed the long Y in Down's syndrome and his related persons. Long Y has also been observed in a Marfan's syndrome (Kallen and Levan, 1962) and in a case of oligospermia (Van Wijck et al., 1962). A consistenly large Y was observed in a case of hypogonadism and in his normal father, brother, son and paternal uncle by Chapelle et al. (1963a,b). They also observed an inconsistently large Y in Down's syndrome, his normal father and brothers. Long Y chromosome has also been observed in a mentally retarded individual and his normal father, in an individual with testicular feminization and his normal father as well as in an azoopermic individual (Van Wijk et al., 1962; Tonomura and Ono, 1963). Dekaban et al., (1963) also observed 15% increase in the length of Y chromosome in a Down's syndrome patient, in his normal father and in a normal father of a mongoloid female. However, an unusally long Y chromosome has also been observed in a normal men by various authors (Hungerford, 1964; Tjio, 1964; Gripenberg, 1964).

Polymorphism of Y chromosome has also been studied in different population of the world. Cohen et al. (1966) studied Y chromosome polymorphism among the White and Non-Jewish males of New York and observed the frequency of long Y to b e 5%. Court Brown (1967) observed a frequency of 1.5% of the long Y among the Scots newborns. A frequency of 13.4% of the long Y was observed among the Finnish (Unnerus et al., 1967). Torre and Gimenez-Martin (1970)

observed a frequency of 18.6% of the long Y carrying individuals among Spanish population. The frequency of the long Y among newborns of New Haven, and negro of New Haven and Negro of New Haven were found to be 6.6%, 14.9% and 14.8%, respectively (Lubs and Ruddle, 1970, 1971). Among the Swedish the frequency of long Y was found to be 9.4 (Lins and Sundequist, 1971). Frequency of the long Y among German children was 2.1% (Zankl and Zang, 1971); among Polish students the frequency was 2.9% (Hubner, 1971); among the new borns of Danes it was 1.4% (Nielsen and Friedrich, 1972); the frequency of the long Y among the Canadian newborns was 0.9% (Hamerton et al., 1972); among the newborns of Danes it was 1.0% (Friedrich and Nielsen, 1973). The frequency of long Y among normal individual of Ontario was 12.2 (Soudek et al., 1973); among the Estonian (Russian) it was 3.8% (Mikelsaar et al., 1973); among the Russian newborns it was 1.5% (Bochkov et al., 1974). Among Indians, Ghosh and Singh (1975) found a long Y chromosome in 5% of the Rajput and 3% of the Punjabi populations. However, during the present investigations the frequency of a long Y chromosome was found to be 5% in the Saini and Kamboj from Haryana population. It may also be noted from the above that geographical as well as racial differences do occur.

The variation in the length of the Y chromosome is probably distributed in populations according to a Gaussion curve (Unnerus et al., 1967). The length of Y in proportion to the average of other small acrocentric chromosomes is usually calculated. This is termed the Y index and illustrates the relation of the Y to those chromosomes. Tonomura and Ono (1963) calculated Y /G index of the Swedish population was found to be 0.734 (Lins and Sundequist, 1971). On the other hand, Y/E index was found to be 0.83 and 0.88 in Danish newborn male children and youth from Danish prison respectively (Nielsen and Henriksen, 1972; Zeuthen and Nielsen ,1973).

During the present investigations it was observed that the values of Y indices were the highest among the Saini and the lowest among the Jat. The t test showed that the difference for Y/D index was found to be statistically non-significant among the ethnic groups studied as well as between the Ahir and Japanese (Table 2). The difference in the Y/E index was found to be statistically non-significant among the Jat and Ahir, the Kamboj and Ror and the Kamboj and Punjabi. The difference for Y/E index was statistically non-significant among the Jat and the Kamboj, the Jat and the Ror and the Kamboj and Ror. The difference for Y/G index was also statistically nonsignificant among the Jat and Kamboj, the Jat and Ror, the Ahir and Kamboj, the Ahir and Ror, the Saini and Kamboj, the Saini and Ror and the Kamboj and Ror (Table 2). The remaining inter-groups comparisions showed significant differences for the various indices.

Various factors have been suggested to be responsible for variation in the length of the Y chromosome. The variation, partly or wholly, may be simply a result of varying degree of contraction of the chromosome during the cell division (Bishop et al., 1962). Gripenberg (1964), however, considered that the increase in length may be the result of addition of chromosomal substance and cited the evidence that the extra long Y chromosome bears two secondary constrictions. From the qualitative analysis using tritium labelled thymidine it was inferred that the difference in the size of the Y chromosome is a morphological feature without any functional significance (Chapelle et al., 1963a, b; Kikuchi and Sandberg, 1965). The long Y chromosome may be a result of structural changes (Wahlström, 1971) and the duplication of finite lenth of chromosome may be the one mean by which the long Y chromosome has evolved (Tishler et al., 1972).

Table 2 : Mean seperation analysis based on 't-test' [those means underlined by the same and dotted lines (only the two means), are not significantly different from each other]

Y/D INDEX

RAJPUT	PUNJABI	JAPANESE	SAINI	KAMBOJ	ROR	AHIR	JAT
0.6504	0.5948	0.5920	0.5340	0.5130	0.4835	0.4800	0.4445

Y/E INDEX

RAJPUT	SWEDISH	FINNISH	PUNJABI	SAINI	ROR	KAMBOJ.	AHIR	JAT
0.7479	0.7340	0.7306	0.6765	0.5700	0.5485	0.5385	0.5180	0.5045

Y/F INDEX

JAPANESE	GERMAN	JEWS	NEGROS	RAJPUT	INDIANS	NON-JEWS	PUNJABI	SAINI	AHIR	ROR	KAMBOJ	JAT
1.0019	0.9856	0.9410	0.9223	0.9019	0.8842	0.8621	0.7971	0.7080	0.6710	0.6495	0.6370	0.6215

Y/G INDEX

JAPANESE	RAJPUT	PUNJABI	SAINI	KAMBOJ	AHIR	ROR	JAT
1.2900	1.1597	1.0272	0.9835	0.9495	0.9490	0.9265	0.9120

Some workers observed that the length difference in the variant Y chromosome involved the highly fluorescent segment only (Bobrow et al., 1971; Robinson and Buckton, 1971; Laberge and Gagne, 1971; Knuutila and Gripenberg, 1972). However, Schnedl (1971) and Soudek et al. (1973) observed that in addition to strongly flourescent portion, weekly fluorescent proximal portion of the long arm also showed modest variation in the length.

Stern et al. (1964) suggested that the Y chromosome is apparently sterile in the sense that no genes are present on this chromosome and the functional morphological variability of the human Y chromosome is not very clear. The selective pressure that maintains the Y chromosome polymorphism may be investigated by correlating Y length with fertility. Patil and Lubs (1977) found that the proportion of abortion was increased two-fold in the matters of long Y infants, compared to others. They also observed that 1.9% of the males under investigation had Y/E index greater than one.

It is interesting that the Y chromosome length varies considerably even in the cells derived from the same individual. (Bender and Gooch, 1961; Makino and Muramoto, 1964; Hungerford, 1964). The existence of two types of cells, one with a normal Y and the other carrying an unusually long Y chromosome was reported by Van Wijck et al., (1962). It has also been reported that the long Y chromosome occurs in normal and healthy males as well as males associated with various congenital disorders (Makino et al., 1964; Takagi et al., 1964; Kato et al., 1965). Makino and Takagi (1965) further concluded that the increased length of the Y chromosome is a heritable charater, other workers also supported this feature (Bishop et al., 1962; Chapelle et al., 1963a, b; Tonomura and Ono, 1963; Dekaban et al., 1965).

However, it remains a question as to whether the length of Y chromosome is inherently liable to vary responding to an exaggerated and non-coordinated spiralization of the Y, or otherwise corresponding to a structural change of chromatin as a result of deletion, duplication, or translocation (Makino and Takagi , 1965).

It is evident from the foregoing discussion that the wide range of variation in the human karyotype is tolerated and the ethnic polymorphism is one of the strongest factors responsible for maintaining this variation. However, much work has to be done in this area before anything conclusive can be said about the mechanism and the way such chromosome polymorphisms are maintained.

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