1. GENERAL INTRODUCTION AND CONTEXT OF STUDY

1.0 Introduction

The objective of the present thesis is to obtain further knowledge about (i) the effects of inbreeding on certain (a) anthropometric (b) psychometric (c) physiometric and (d) dermatoglyphic variations in a hitherto unstudied human population; the Qureshi Muslims of Aligarh city, Uttar Pradesh and (ii) the light they shed on the genetics of the traits studied.

It is based on an intensive field investigation in the population for a period of about three years followed by critical analysis and interpretation of the data in the laboratory of the Aligarh Muslim University and the University of Calcutta for about two years.

1.1 Concept of Consanguinity and Inbreeding

Preferential mating between relatives is an important deviation from the model of Panmixis. Individuals who are related through one or more common biological ancestors are called consanguineous relatives. In human population marriage largely regulates mating (Mukherjee, 1971) and consanguineous marriage thus leads to inbreeding. In a strict sense, however, inbreeding occurs in the offspring of consanguineous parents. Concisely inbreeding is the genetical consequences of consanguineous matings.
1.2 Muslim Traditions about Marriages

The Muslim populations all over the world have generally been found to practise consanguineous marriages between first cousins and more distant relatives. However, according to the precepts of the Holy Quran*, a Muslim male is prohibited to have incestuous marriage between first degree relatives of the following types: mothers, daughters, sisters, foster mother (who gave suckle), foster sisters, wives' mothers.

1.3 Reasons for Consanguineous Marriages

The reasons for consanguinity are suggested to be for economic and social purposes. In the studies done in Andhra Pradesh (Dronamraju and Meera Khan, 1963a), five reasons are reported for preferring consanguineous marriages: (a) to keep the cultivable land in larger subdivisions for growing food crops such as rice, (b) parental influence, (c) mutual knowledge of families, (d) economic benefit, & (e) the extreme youth of the brides. According to Centerwall and Centerwall (1966) it is practised in order to minimise the amounts of dowry to be given in the same social status.

Beyond these reasons, among the Muslim groups, the marriage is influenced by religious injunctions. Another reason for these unions is to maintain the purity of blood. Besides this, the other fact is to retain the family

* The Holy Quran; IV, 23
property as in Islam the girls are owner of the fixed portion from the landed family property. Further, it is easier to select an eligible girl from one's own family as compared to others.

1.4 Types of Consanguinity

Consanguineous marriages are of many types (Morton, 1961). The children, having same parent are called 'sibs'; their children are the first cousins and their grand children are the second cousins, children with one common parent are half-sibs. The child of one's first cousin is called as one's "first cousin once removed" and the grand child of one's first cousin is one's "first cousin twice removed". Thus many combinations of relationships between spouses are theoretically possible. Different types of consanguineous unions are shown in pedigrees (Plate 1). The closer the consanguineous relationship between parents, more intense is the degree of inbreeding or of autozygosity.

1.5 Frequency of Consanguineous Marriages

Consanguineous marriages are practised to a greater or lesser extent among most of the religious and ethnic groups of the world. However, the rate of consanguinity differs widely due to various socio-economic and cultural factors. In India, the North Indians largely shun this type of marriage, whereas the South Indians of Dravidian origin practise even uncle-niece or aunt-nephew marriages
(Sanghvi, 1966; Mukherjee, 1972, 1992). The rate of consanguinity varies on factors like (a) state (b) religion (c) caste (d) tribe (e) language (f) occupation (g) socio-economic stratum (h) education (i) residence (j) isolation (k) population size etc. (Rao, 1983).

Over all, consanguinity is practised mainly due to lack of social mobility and is prominent among socio-religious isolates, the neoconverts and also groups with primitive traditions. However, a secular trend in lowering the consanguinity has been seen for many of the populations the world over (Imaizumi and Shinozaki, 1984).

However, in the transitional phase the decline in the frequency of consanguineous marriages is not reflected in the change of inbreeding coefficient (Mukherjee, 1992).

1.6 Coefficient of Inbreeding

The offspring from the consanguineous marriages are called as inbred ones. The degree of inbreeding is measured by the coefficient of inbreeding. Two major formulations have been proposed for the coefficient of inbreeding. It is defined as (i) the probability of two homologous alleles of an individual being identical by descent (Haldane and Moshinsky, 1939; cotterman, 1940; Malecot, 1948), (ii) the genetic correlation between uniting gametes (Wright, 1921a; Wahlund, 1928).

The coefficient also measures the proportion of autozygous alleles (Mukherjee, 1984).
The coefficient of inbreeding is conventionally designated by a symbol 'F' introduced by Wright, which is proportional to the reduction in heterozygosity due to inbreeding in a base population. The \( F_i \) for an individual is calculated by using the formula:

\[
F_i = \frac{1}{2}^n (1 + F_a)
\]

where \( n \) is the number of persons along the path through a common ancestor connecting two parental gametes and \( F_a \) the inbreeding coefficient of that common ancestor (Mukherjee, 1984).

1.7 Genetical Significance of Inbreeding Effects

1.7.1 Homozygosis

Several studies have been done from time to time to note the effects of parental consanguinity. Parental consanguinity increases the frequency of homozygotes in the offspring at the expense of heterozygotes, recessive and additive phenotypes would thus increase in frequency in the inbred individuals. This is true for single gene traits as well as multifactorial traits. In other words, inbreeding brings out previously hidden recessive and additive alleles contributing to the phenotypic variance (Crow and Kimura, 1970) and hence it adds to the genetic variability (Jensen, 1978, 1983). Thus, phenotypic manifestation of complex and quantitative biological variations contribute significantly to the understanding of the genetics of those traits when
other sources of variation such as environmental variations are largely controlled. Such manifestation of the anthropometric, psychometric, physiometric and dermatoglyphic traits in inbred individuals are considered in the present thesis.

1.7.2 Recessive and Additive Genes

A carrier of a rare gene is unlikely to marry another carrier if mating is random, but this is more likely to happen if he marries a consanguineous relative. The proportion of the affected person in the population due to inbreeding is

\[
\frac{\alpha}{q(1-q) + \alpha} = \frac{\alpha}{q + \alpha}
\]

where, \( q \) = frequency of rare gene in the given population, \( \alpha \) = frequency of the affected persons in the population in the absence of inbreeding. Dahlberg (1948) made these considerations on the basis of an estimate of the recessive gene frequency.

\[
K = \frac{C (15q + 1)}{(C + 16q - qC)} \quad \text{and} \quad q = \frac{C (1 - K)}{(16K - 15C - CK)}
\]

where \( K \) = proportion of affected persons whose parents are first cousins. Therefore, the study of inbreeding has been useful in detecting rare recessive genes.
A limited number of studies on inbreeding effects on quantitative traits in man have been conducted. So, much scope of discovering genetic backgrounds of physical, physiological and behavioural characters and also their genotypic variations exist and, where possible, it leads to identification of the homozygotes for recessive or additive genes. The present investigation is concentrated onto these problems.

1.7.3 Morbidity and Mortality

Due to increased homozygosity of recessive genes in the inbred individuals, morbidity and mortality have been found to increase with increase of inbreeding. Increased prenatal, infant, child and adult mortality and also morbidity causes enhanced selection with inbreeding (Elderton, 1911; Morton and Hussels, 1970; Mukherjee, 1974 1984; Mukherjee and Bhasker, 1974; Rao and Mukherjee, 1975; Rao and Inbaraj, 1977b; Afzal, 1984; Chitty and Winter, 1989; Gillies et al., 1984; Reddy 1992).

1.7.4 Fertility and Sterility

Divergent views are held by various investigators who have explored the relationship of inbreeding to fertility and sterility. Some authors (Bemiss, 1958; Arner, 1908; Slatis et al., 1958), conclude that inbreeding does not influence fertility while increased infertility can be seen (Eaton and Mayer, 1954; Cross and Mckusick, 1970). The proportion of sterile females increased in consanguineous
marriages (Sutter and Tabah, 1952; Book, 1957; Sjöström et al., 1958). But this phenomenon is not directly correlated with the degree of inbreeding or the deliberate use of birth control measures (Sutter, 1958; Schull, 1959). In general, the higher fertility rates have been reported among females in consanguineous marriages than non-consanguineous in Hiroshima and Nagasaki (Schull and Neel, 1965); Hirado (Schull et al., 1970); Egypt (Hussien, 1971); Brazil (Freire-Maia and Azevedo, 1971); India (Reid, 1973; Mukherjee, 1974, 1984; Mukherjee et al., 1977; Afzal and Sinha, 1984; Afzal, 1984). This high fertility may reflect the role of reproductive compensation (Schull et al., 1970; Reed, 1971; Mukherjee et al., 1977) or a difference of interspouses' relationship between the consanguineous and non-consanguineous individuals (Reid, 1973).

1.7.5 Genetic Load

Studies on consanguineous marriages indicate that most individuals in a population are heterozygous for one or more of the recessive genes which would become homozygous, and would cause severe impairment or death with inbreeding. One measure to assess the magnitude of morbidity and mortality of the gene pool of a population is known as genetic load (Morton, 1982). There are various forms of genetic loads, such as expressed genetic load, total genetic load, segregational load and mutational load. Consangunity thus adds to the segregational load. The inbreeding data from human
population have been used to estimate the average number of harmful genes per individual by several authors (Slatis, 1954; Penrose, 1957). The estimates of heterozygous deleterious genes per zygote calculated from consanguinity effect, range from 2 to 9 (Scott Emuakpor, 1974). They have been fitted on a linear regression equation to the negative logarithm of the surviving proportion of the inbred for $P$:

$$-\log P_s = A + BF$$

1.7.6 Selection Intensity

Mutations are generally detrimental and dominant mutations are eliminated rapidly whereas harmful recessive mutations accumulate in the population (Stern, 1960; Bodmer and Cavalli-Sforza, 1976). Inbreeding thus leads to increased selection intensity by furthering the elimination of recessive harmful genes.

The practice of repeated inbreeding through generations may cause the decline of harmful genes as suggested by Sanghvi (1966) through mathematical logic on the assumption of no fresh mutation and absolute genetic isolation of population. The condition, however, may not strictly prevail in human population.

However, several Indian studies have confirmed that the frequency of malformation and mortality rates increase in inbred groups of any endogamous population (Dronamraju and Meera Khan, 1963b; Centerwall and Centerwall, 1966; Kumar
et al., 1967; Chakravarti, 1969; Jacob and Jayaball, 1971; Murthy and Jamil, 1972; Reid, 1973; Mukherjee, 1971, 1974; Mukherjee et al., 1977; Mukherjee and Bhasker, 1974; Rao and Mukherjee, 1975; Basu, 1975; Ghose and Majumder, 1978; Reddy and Rao, 1978; Reddy, 1985; Rao and Inbaraj, 1979; Ansari and Sinha, 1983), although this increase is not always significant (Notani et al., 1968; Arner, 1908; Slatis et al., 1958).

1.8 Studies on Consanguinity

Theoretical and empirical studies on inbreeding are available from different parts of the world, and for different human populations since long back. Direct experimental studies are not possible among humans, however, these are widely reported in plant and animal literature. These studies are concerned with various aspects of inbreeding on a smaller or larger scale. On the whole the following trends can be traced in the general survey of literature.

1.8.1 Outside India

Theoretical works are accredited to mathematicians of the evolutionary school (Fish, 1914; Jennings, 1916; Wright 1931, 1951; Haldane and Moschnisky, 1939; Cotterman, 1940; Malecot, 1948; Li, 1955; Falconer, 1960; Crow, 1954; Kimura and Crow, 1963). Survey on the levels of consanguinity are available from various countries of Europe viz., France
(Boudin, 1962; Wultz, 1925), Scotland (Mitchel, 1965), England and Wales (Shield and Slater, 1956; Bundey et al., 1990), Germany (Mulhal, 1892), Italy (Cavalli-Sforza, 1956), Ireland (Cameron, 1883), Austria (Orel, 1932), Sweden (Fraccaro, 1956), Switzerland (Ruepp, 1935), Denmark (Kemp, 1950), Holland (Sutter and Tabah, 1948) Spain (Pinto-Cisternas et al., 1985), Northern Ireland (Kilpatrick et al., 1955), Japan (Neel et al., 1949; Schull, 1953), U.S.A. (Arner, 1908; Estarbrook and McDougle, 1926; Woolf et al., 1956; Brown, 1951), Canada (Miner, 1939), Argentina and Uruguay (Friere-Maia, 1957b), Brazil (Friere-Maia, 1952, 1954, 1957a), Jamaica (Davenport and Steggerda, 1929, Doran, 1942). The literature is extensively reviewed in Friera-Maia (1982) and more recently by McCullough and Rourke (1986) pointing toward highest local rates of inbreeding in Brazil, Japan, Israel (Freundich and Hino, 1984) and India, with lower frequencies in the U.S.A., and European countries. Studies have been done in Venezuela (Pineda et al., 1985).

Muslim populations in third world countries have been covered more recently viz., Kuwait (Al-Awadi et al., 1985), Turkey (Basaran et al., 1988), Sudan (Saha and Shaikh, 1988), Egypt (Hafez et al., 1983), Lebanon (Kalostian et al., 1980) and Pakistan (Shami and Siddiqui, 1984; Shami and Mahmood, 1986; Shami, 1985). Some data are available on the effects of incestuous unions in the west, namely U.S.A. (Adams and Neel, 1967; Adams et al., 1967; Carter, 1967; Seemanova, 1971).
1.8.2 India

Intensive investigations on various rural populations have been done in India also, in the Maharashtra and Andhra Pradesh (Sanghvi, et al., 1956; Sanghvi, 1966; Dronamraju and Meera Khan, 1961a,b; Dronamraju, 1964, 1967; Stevenson et al., 1966; Ali, 1968; Veer Raju, 1973; Srinivasan and Mukherjee, 1976; Roy Chowdhery, 1976; Rao, 1978, Rao et al., 1971, 1972; Rao and Inbaraj, 1977a,b, 1980). Reviews of the results of such studies have also been attempted (Sanghvi, 1966; Mahapatra, 1966; Chakravarti, 1968; Badaruddoza and Afzal, 1992). Studies on morbidity and clinical aspects were made by Verma (1980), Devi et al., (1987), Sinclair (1972), Joshua (1974), Centerwall and Centerwall (1966). Reports are also available from Central India (Goswami, 1970), Northern and Eastern parts and also among the various caste and tribes (Karve, 1957; Yadav, 1968; Ghosh, 1972; Malhotra, 1967; Mukherjee, 1974).

1.8.3 Indian Muslims

Studies among Muslims have been scarce, though muslims practise inbreeding in India and abroad. Reports are available from Northern and Eastern parts of India mainly Uttar Pradesh and Delhi (Basu and Roy, 1972; Basu, 1978; Afzal and Badaruddoza, 1990; Badaruddoza, 1990), Kashmir (Kashyap, 1978), Bihar (Ansari, 1980; Afzal and Sinha, 1983a,b, 1984), West Bengal (Barua, 1976; Haq, 1976, Mukherjee, 1992), Rajasthan (Basu, 1975) among Shia, Sunni,
Ahmadia and of different biradaris, mainly Sayyad, Pathan, Shaikh, Bohras, Ansari etc. Studies in Madhya Pradesh (Goswami, 1970), Kerala (Ali 1968), Tamil Nadu (Rao and Inbaraj, 1977a,b), Andhra Pradesh (Sanghvi, 1966; Mukherjee and Bhasker, 1974) have been done along with other communities.

1.9. Studies on Inbreeding Effects on Quantitative Traits

Quantitative genetics in man has been mainly confined (i) to the estimate of components of variation due to additive gene heritability, non-additive genes and environmental influence and (ii) to the identification of segregation at single loci (Harris and Kalmus, 1949, Harris, 1966; Mukherjee and Rao, 1976). Most of the studies are based on correlation between relatives. The high correlation only suggests but does not confirm strong genetical influence on a trait (Murphy, 1979; Mukherjee, 1984).

The analysis based on correlation does not throw light on the genotypes, positive or negative effect of genes and dominant/recessive epistasis between loci (Mukherjee, 1984). Even the new application of path analysis (Bock, 1979; Rice et al., 1990; Roberts et al., 1978) fails to throw light on such phenomenon. Therefore, inbreeding effect on quantitative traits provide yet another approach to study these processes.

1.9.1 Inbreeding Depression

The most striking consequence of inbreeding is the
reduction of the mean values shown by characters connected with reproductive capacity, reduction of body size, delay of development or functional impairment known as inbreeding depression. The inbreeding depression had first been observed in plants (Jenning, 1916; Jones, 1924) and animals (Falconer, 1960, Pirchner, 1969). The characters that form an important component of fitness, such as litter size, lactation in mammals, egg number in poultry show a sensitivity to inbreeding (Falconer, 1960). Significant inbreeding depression of particular traits has been obtained in Japanese children and adults among the Japanese Hutterites, Egyptian Nubians, Italians and Brazilian immigrants. The heights of French children also decline in areas in which the inbreeding coefficient (F) is high.

1.9.2 Elevation of Means

Certain quantitative characters show a consistent increase of mean under inbreeding such as fat content of milk in cattle, egg size of poultry, age at menarche in girls, systolic and diastolic blood pressure in human beings (Table II). Recognition of change of mean on either direction as a result of inbreeding is important for using these results for study of genetics of the traits.

1.9.3 Heterosis

Complementary to the phenomenon of inbreeding depression is its opposite, 'hybrid vigour' or heterosis. When inbred
lines are crossed, the progeny shows an increase of those characters that previously suffered a reduction from inbreeding. The fitness lost on inbreeding tends to be restored on crossing. Heterosis is thus defined as inbreeding depression in reverse (Falconer, 1960). Heterosis may be observed mostly in farming such as egg production in the poultry and growth in pigs (Nordskog and Ghostley, 1964).

Thus heterosis, just like inbreeding depression, depends for its occurrence on dominance. Loci without dominance \((d = 0)\) cause neither inbreeding depression nor heterosis. The amount of heterosis following a cross between two particular lines or populations depends on the square of the difference of gene frequency between the population. If the population crossed do not differ in gene frequency, there will be no heterosis and the heterosis will be greatest when one allele is fixed in one population and other in the other population. When we consider the joint effect of all loci together, so far as the genotypic values are attributable to the separate loci, they combine additively. If there is no change of mean in the inbred group, it may mean absence of dominance of genes involved in the traits. If some loci are dominant in one direction and some in the other, their effects will tend to cancel each other, and no heterosis may be observed, in spite of the dominance at the individual loci. Therefore, the occurrence
of heterosis depends on directional dominance and the absence of heterosis is not sufficient ground for concluding that the individual loci show no dominance.

1.10 Theoretical Models

1.10.1 Genetical Effects of Inbreeding on Means

The basic model for genetical analysis of quantitative traits with known inbreeding levels is contained in the following deduction (Mather, 1949, Dickinson and Jinks, 1956 Falconer, 1960).

\[ m_F = m - 2F \sum d_p q \]

where \( m \) and \( m_F \) are mean values in the absence and presence of inbreeding at the level \( F \) respectively, \( p \) and \( q \) are allele frequencies \( (p + q = 1) \) and \( d \) the phenotypic value of heterozygote measured from the mid-value between two homozygotes at a locus, and \( \sum \) denotes summation over the involved loci assumed to be mutually additive. Thus, the change of mean would reflect an amount of overall dominance of genes affecting the trait \( \sum d \neq 0 \). Secondly, as the change of mean would be in the opposite direction of the dominant phenotype, it would indicate whether the recessive genes should have positive or negative effects. If the genes having positive effects (which increase the value) are dominant, \( d > 0 \), over their alleles which reduce the value, inbreeding results in a depression of mean. If positive alleles are recessive, that is \( d \) has a negative value, the
mean value is elevated by inbreeding.

Therefore the convention of searching for presence or absence of inbreeding depression is not so comprehensive as is the search for change of means in either direction, depression or elevation.

1.10.2 Linear Effect

The inbreeding effects on mean are expected to be linear with the inbreeding coefficient $F$. So that, the change of mean should be on a straightline when plotted against $F$ (Wright, 1951; Morton, 1958). The effect of the trait is as follows:

$$M_I = a (p - q)$$

$$-2 \Sigma dpq = M_O - M_I$$

where $M_I$ is the mean of homozygous population ($F=1$). Thus, $M_F = M_O + (M_O - M_I)F$ where $0, I, F$ denote no inbreeding, complete inbreeding and intermediate inbreeding respectively. The expression $(M_O - M_I)$ is the coefficient of linear regression of the quantitative traits on $F$.

1.10.3 Non-linear Effect Due to Epistasis

If, there is dominance of some genes at different loci and if there is epistasis between the loci, such as there is some dominance x dominance deviation, the mean value of the inbred series of the population will be related with the inbreeding coefficient in a non-linear manner (Kempthorne, 1957), who also derives that in a random mating population with known inbreeding coefficient $F$ among inbred groups, the mean $m_F$ of this group, in the absence of selection, will be
given by the formula:

\[ M_F = M_0 + FD_1 + F^2D_2 + F^3D_3 + \ldots \]

where, \( M_0 \) is the original mean of the randomly bred population and \( D_1, D_2, D_3, \ldots \) are dominance deviations.

Crow and Kimura (1970) again clarify that if there is an interaction of dominance of one locus with the dominance of another locus (epistasis between two dominance) there will be a diminishing epistasis with inbreeding. This indicates that the distribution of mean measurements in relation to the inbreeding coefficient is concave upwards as a result of positive interaction between the dominant genes at multiple loci. The interaction between the dominant genes would gradually decrease with the increase of homozygous recessive genotypes.

\[ M_F = M_0 - DF + EF^2 \]

where \( M_F \) is the mean of the population with \( F, D \) and \( E \) are the total dominance and total epistatic effects respectively. Therefore, when \( E = 0 \) (no epistasis) the inbreeding change is linear on \( F \), when \( E > 0 \) the curve of depression with \( F \) would be concave upwards. In conclusion, if there is epistatic interaction between loci, the relationship between the mean and the inbreeding coefficient is not linear. The non-linearity is due to the interaction deviation of double or multiple heterozygotes. The frequency of double heterozygotes declines in proportion to \( F^2 \). Therefore as \( F \) increases,
the rate of depression of the mean increases, if the interaction deviations are on average positive i.e., favourable, and the rate decreases if they are negative. So the interaction affects the linearity, and epistasis without dominance cannot itself cause any inbreeding depression. Increasing the homozygosity for loci would reduce the inbred means by less than sum of their individual effects. If $E < 0$, when $D > 0$, the epistasis is reinforcing and the inbreeding depression is greater than cumulative. So this non-additive epistasis between dominant genes of different loci would give rise to an exponential curve of means for different $F$ values. In the present thesis both linear and non-linear components of the change of means on inbreeding have been investigated.

1.10.4 Effect on Variance

As a result of inbreeding, genetic differences between homozygotes for different alleles become larger whereas the total genetic variance within the lines decreases. Similarly the total additive genetic variance increases.

1.10.5 Additive Variance

The total additive variance in the whole population is the sum of the within line and between line components, and is equal to $(1 + F)$ times the original variance. Therefore the additive variance in a degree of inbreeding $F$, i.e., $V_F$, increases by an amount equal to $F$ (Wright, 1921b; Li, 1955;
Kempthorne, 1957; Falconer, 1960).

\[ V_F = 2\sum pqa^2 (1+F) = V_A (1+F) \]

where \( a \) is the genotypic value of the positive homozygote and \( V_A \) the additive variance in the population with \( F = 0 \), subsequently when inbreeding is complete the additive variance in the population as a whole is doubled and all of it appears as the between line component. So there would be a linear increase of the additive genetic variance on inbreeding.

### 1.10.6 Dominance Variance

The changes in the components of variance arising from additive genes will have to be seen to be independent of the gene frequency in the base population and this would make the prediction of inbreeding effects on such variance difficult. The additive proportions of genetic variance for different degrees of dominance and gene frequencies show that the effect of dominance on genotypic variance is serious for the case of fully dominant genes, when recessive allele is at low frequency (Li, 1955). The within-line variance at first increases, reaching a maximum when the coefficient of inbreeding is a little under 0.5 as it remains at fairly high level until the coefficient of inbreeding approaches 1.0 (Robertson, 1952). Reid (1973) demonstrated that:

\[ V_F = \sum a^2 [4pq + H (2 -4p - H)] \]
where \( a \) is the genotypic value of the positive homozygote, 
\[ H = 2 (pq - F_{pq}) \] and \( p \) is the frequency of dominant allele 
at the locus. This equation represents the multilocus system 
of additive epistasis. Therefore on average, the variance 
within the lines will increase in the early stages of 
inbreeding. This increase of variance would be detectable 
in practice only if a substantial part of the genetic 
variance were due to recessive genes at low frequencies.

1.10.7 Environmental Variance

Environmental component of variance may differ 
according to the genotype, in particular, that inbred 
individuals often show more environmental variation than 
non-inbred individuals. Any differences of phenotypic 
variance between highly inbred lines and the \( F_I \) between them 
(hybrid) must be attributed to a difference of the environ-
mental component, because the genetic variance is 
negligible in amount among the hybrids as well as in the 
Therefore, phenotypic variance, \( V_P \) is not reflecting the 
genotypic variance, \( V_G \). But the estimates of change in \( V_G \) 
can be based on systematic change in \( V_P \) (Hogben, 1933) if 
the environmental variance \( V_E \) is independent of the genotype 
and \( F \). Subsequently, in the genetically mixed group, the 
phenotypic variance follows the equation: 
\[ V_P = V_G + V_E + 2COV_{GE}. \] 
Subtraction of the two phenotypic variances, therefore,
yields an estimate of \( V_G + 2COV_{GB} \). Thus, neglect of a correlation between genotype and environment leads to the covariance being included with genotypic variance. However, animal experiments reveal the same, because the environment is a direct consequence of the genotypic value (Robertson and Reeve, 1952; Mather, 1953; Waddington, 1957). But this hypothesis is less satisfactory with human intelligence because the environmental effects on the children are not a consequence of their own genotypes but of their parents' genotypes.

Wolanski (1974) assumes that the heterozygotes is to be more eco-sensitive in humans. This hypothesis suggests a greater environmental variation within the non-inbred series of individuals than within the inbred lines. But these are often merely hypothetical and not yet precisely determinable in human or wild populations.

1.11 Studies on Humans

Studies on inbreeding effect on physical, physiological and psychological measurements have been conducted over the last three and half decades. But the limitations of the data, the objectives and interpretation in many earlier studies, prevent us from arriving at any firm conclusion about the genetical implication of their results, as pointed out by Mukherjee (1984).

In all, earlier studies, specially from outside India,
suffered several limitations like biased sample as conscripts, mixed populations, non-control of gene pool and environmental variations, unrepresentative and unequal samples from different degrees of inbreeding. In the previous studies, most of the investigators looked for presence or absence of inbreeding effect instead of looking for the type of effects. They only considered inbreeding effects in depression of mean values. In fact inbreeding will naturally lead to homozygosity but in some cases as in the case of vital characters like certain enzymes (Jenkins et al., 1985), the progress of homozygosis is disrupted by selection (Mukherjee 1990b). As mentioned earlier that the change of means is not essential for the effects of inbreeding, because there may be homozygous additive genes. This should not be interpreted as absence of inbreeding effects. As the characters selected, like physical measurements, are believed to be related to socio-economic and nutritional conditions, sometimes, effects of depression or lowering of means, are sweepingly interpreted as effects of environmental factors rather than due to homozygosity.

In fact positive results have been obtained by critical studies avoiding or minimizing such limitation in a few Indian populations (Lakshmanudu, 1980; Mukherjee, 1984; Mukherjee, 1990a). The inbreeding effects in a few Indian studies have been examined also in terms of change in distribution and variations.
Therefore, there is urgent need for extending such studies to other populations and to further improve the sampling design if possible. Large samples are needed to get the significant difference between the groups, but at the same time, large samples are unrealistic. Because the familial aggregation of consanguineous marriages makes only a few clusters of family of inbred and non-inbred from the same genetic background which leads to demic subdivision within the population. This makes it different to obtain adequate samples of each level of inbreeding. Other studied Indian samples have not also been free from this problem due to the nature of the population. Therefore, there is a need for specific population where the consanguineous marriages are relatively more uniformly distributed than concentrated in small section of the population. In the present study, it has been possible to find out urban population with uniformly distributed and largely homogeneous with one socio-economic status and genetic background.

Studies of inbreeding effects on quantitative, specially anthropometric, psychometric, physiometric, dermatoglyphic traits are only a few in India as elsewhere (Mukherjee et al., 1977; Malhotra, 1979; Lakshmanudu, 1980; Rao and Inbaraj, 1980; Mukherjee, 1984; Agrawal et al., 1984; Basu and Jindal, 1987; Afzal, 1988; Badaruddoza, 1990) This thesis provides a critical examination of the entire evidence of some aspects of inbreeding effects on quantitative
biological variations in humans, including an illustration through our studies in India. This thesis will further the possibilities of utilizing the data for the genetic analysis of quantitative traits viz., anthropometric measurements; intelligence quotient; blood pressures and also dermatoglyphics.

The approach for analysis of present data can also baffle up the conventional techniques. Because all previous studies are chiefly concerned with the change of means of physical, psychological and physiological measurements (Table I, II) on inbreeding.

1.12 Dermatoglyphic Genetics and Inbreeding

That dermatoglyphic variations are not influenced by environmental factors other than in utero conditions is amply demonstrated by embryological studies (Cummins and Midlo, 1943), repeated examinations on same individuals, initiated by Herschel, (1880) (cited in Cummins and Midlo, 1943), and even studies on Egyptian population (Penrose cited in Mukhejee, 1980). The genetic background of various aspects of the patterns were established by biometric studies by Penrose (1954); Holt (1960); Mukherjee (1966, 1967) and others.

Knowledge of inbreeding effects on dermatoglyphics is rare in India and is so far limited to few Indian studies (Sabarni and Kukherjee, 1975, Mukherjee et al., 1980; Mukherjee, 1989, 1990a,b) in a number of populations from
different parts of India and provide consistent results. These results, suggesting homozygosity of extreme pattern intensities on fingers, also agree with results on studies on the effects of village endogamy, which shows indirect effects of inbreeding (Marquer and Jakobi, 1976; Mukherjee and Chakravarti, 1964; Loesch, 1971; Roberts and Coope, 1972). This suggests the usefulness of inbreeding studies on dermatoglyphics in future advancement of the genetics of dermatoglyphic traits.

Besides, there is a critical importance of the studies of dermatoglyphic effects of inbreeding on quantitative characters in general. The observed difference between inbred and non-inbred samples in several earlier data from non-Indian populations failed to convince many workers about the genetical significance of those results due to concomitant socio-economic and other environmental factors including age. To bring home the genetic significance of the homozygous effects, dermatoglyphic studies on inbred and non-inbred samples have been found to be convenient. The idea that the characters with lower heritability showed greater inbreeding effect only in respect of the change of mean (depression) has to be placed in this context of average dominance/recessiveness of the concerned genes.

The dermatoglyphic results of inbreeding observed so far have certain problems, in view of the limitation of adequate samples of individuals with same F-values. The
change of means which though consistent have only small amount of significance i.e., which may have lower than even 5% probability. The present data have advantage of four precise degrees of inbreeding.

1.13 Limitation of Data

The present analysis of the data shows that inbreeding adversely affects the overall physical, psychological, physiological traits. However, some limitation to our study is to be noticed. It is assumed that all traits studied, except dermatoglyphic characters, are somewhat influenced by environmental factors (Cavalli-Sforza and Bodmer, 1971), although the investigator has taken care to control the socio-economic differences to a large extent. Another difficulty of the present study is that the sample size is not as large as would have been necessary to obtain significant effects of increased inbreeding. This is because the expected amount of change in means and variations on inbreeding among humans are small. Secondly, the samples cannot be enlarged indefinitely because the samples of different levels of inbreeding have to be collected from the same gene pool and similar environmental background, as far as possible, although in the selected population, consanguineous marriages are relatively more uniformly distributed than in the others so far studied, and it limits the population size and availability of suitable samples. The rules of strict endogamy get reinforced by occupational
similarities. Furthermore, since the study aims at obtaining about equal samples from each level (F) of inbreeding from six successive annual age groups in the growing age and in the two sexes. It has not been possible to have a large sample of each category in each group. This limitation has been, to some extent, further removed by pooling the different age group samples using 'z' -scores.

The present study also suffers from a lack of data from highly inbred (F > 0.0625) groups which have not been apparently found in the population. Significant changes of mean on inbreeding are observed in the offspring of the first cousins only, in comparison to first cousins once removed and second cousins.

Interestingly most of the previous studies have not considered the possibility of non-linear effects of inbreeding as well as the natural selection in inbreeding studies in humans except in only few studies in India (Mukherjee, 1984). Therefore, it may be assumed that, in some cases, opposite results of inbreeding specially for blood pressures, can be likely found from increased selection. The other limitation is the difficulty in tracing individuals with low (F<0.0156) inbreeding coefficient, due to non-availability of distant relatives and lack of knowledge of one's own family history by the subjects. So far in the present study the inbreeding classes are restricted to the offspring of only first cousins, first cousins once removed and second cousins. Other types of consanguineous marriages are also very few among Muslims in general because of the presence for close consanguineous marriages.
PLATE - 1. COEFFICIENTS OF INBREEDING FOR P DUE TO VARIOUS TYPES OF MATINGS.
Table - (I)

Quantitative traits in man studied for inbreeding effects  
(After Mukherjee, 1984)

A. Anthropological:

<table>
<thead>
<tr>
<th>No.</th>
<th>Trait</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Stature</td>
</tr>
<tr>
<td>2</td>
<td>Weight</td>
</tr>
<tr>
<td>3</td>
<td>Birth weight</td>
</tr>
<tr>
<td>4</td>
<td>Sitting height</td>
</tr>
<tr>
<td>5</td>
<td>Lower limb length</td>
</tr>
<tr>
<td>6</td>
<td>Head Breadth</td>
</tr>
<tr>
<td>7</td>
<td>Head length</td>
</tr>
<tr>
<td>8</td>
<td>Head girth</td>
</tr>
<tr>
<td>9</td>
<td>Chest girth</td>
</tr>
<tr>
<td>10</td>
<td>Head height</td>
</tr>
<tr>
<td>11</td>
<td>Arm span</td>
</tr>
<tr>
<td>12</td>
<td>Knee height</td>
</tr>
<tr>
<td>13</td>
<td>Tooth diameter</td>
</tr>
<tr>
<td>14</td>
<td>Min frontal breadth</td>
</tr>
<tr>
<td>15</td>
<td>Bisygomatic breadth</td>
</tr>
<tr>
<td>16</td>
<td>Bigonial breadth</td>
</tr>
<tr>
<td>17</td>
<td>Nasal breadth</td>
</tr>
<tr>
<td>18</td>
<td>Nasal height</td>
</tr>
<tr>
<td>19</td>
<td>Biacromial breadth</td>
</tr>
<tr>
<td>20</td>
<td>Bieristal breadth</td>
</tr>
<tr>
<td>21</td>
<td>Morphological facial height</td>
</tr>
<tr>
<td>22</td>
<td>Upper limb length</td>
</tr>
<tr>
<td>23</td>
<td>Cubit length</td>
</tr>
<tr>
<td>24</td>
<td>Bredth of thorax</td>
</tr>
<tr>
<td>25</td>
<td>Depth of thorax</td>
</tr>
<tr>
<td>26</td>
<td>Bistyloid breadth</td>
</tr>
<tr>
<td>27</td>
<td>Max. circ. Upper arm</td>
</tr>
<tr>
<td>28</td>
<td>Calf circumference</td>
</tr>
<tr>
<td>29</td>
<td>Skinfold thickness</td>
</tr>
<tr>
<td>30</td>
<td>Hand breadth</td>
</tr>
<tr>
<td>31</td>
<td>Hand length</td>
</tr>
<tr>
<td>32</td>
<td>Hand index</td>
</tr>
<tr>
<td>33</td>
<td>Cormic index</td>
</tr>
<tr>
<td>34</td>
<td>Cephalic index</td>
</tr>
<tr>
<td>35</td>
<td>Nasal index</td>
</tr>
</tbody>
</table>

B. Somatoscopical

1. Skin colour grade

C. Physiometrical and Psychometrical

1. Blood Pressure-diastolic
2. Blood Pressure - systolic
3. Grip strength
4. Tapping rate
5. Pulse rate
6. Haematocrit
7. Menarcheal age
8. Age when walked
9. Age when talked
10. Length of gestation
11. Intelligence score
12. School grade
13. Social Studies
14.50 metre run
15. Temperament
16. Mazes
17. Colour trail

D. Dermatoglyphical

1. Pattern intensity on specific fingers
2. Pattern intensity on specific palmer areas
3. Triradial number on finger
4. Triradial number on palms
5. Total finger ridge - count
6. ab ridge - count on palms.
Table (II)

Previous studies on inbreeding effects on quantitative traits in Children (After, Mukherjee, 1984.) (Symbols from table I).

<table>
<thead>
<tr>
<th>References</th>
<th>Population</th>
<th>n</th>
<th>Decreases</th>
<th></th>
<th>Increases</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>Significant</td>
<td>Nonsignificant</td>
<td>Significant</td>
</tr>
<tr>
<td>Ichiba, (1953)</td>
<td>Japanese</td>
<td>2829</td>
<td></td>
<td>A.4,6,9,11,24,25</td>
<td></td>
</tr>
<tr>
<td>(Schull, 1958)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Shiroyama, 1953</td>
<td></td>
<td>979</td>
<td></td>
<td>A1, 2, 3,10</td>
<td></td>
</tr>
<tr>
<td>(Schull, 1958)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Morton, 1958</td>
<td></td>
<td>75160</td>
<td>A.1,2,3</td>
<td>A.10</td>
<td></td>
</tr>
<tr>
<td>Furusho, 1961</td>
<td></td>
<td></td>
<td></td>
<td>A.1</td>
<td></td>
</tr>
<tr>
<td>(Morton et.al., 1967)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Slatis and Hoene, 1961</td>
<td></td>
<td>159</td>
<td></td>
<td>A.3,C.11</td>
<td></td>
</tr>
<tr>
<td>Schull, 1962</td>
<td></td>
<td>151</td>
<td>A.7,9,C.4,</td>
<td>A.1,2,4,16</td>
<td>C.17</td>
</tr>
<tr>
<td>Schull and Neel, 1965</td>
<td></td>
<td>353f</td>
<td>A.1</td>
<td>A.2,4,10,12</td>
<td></td>
</tr>
<tr>
<td>Komai, 1963</td>
<td></td>
<td></td>
<td></td>
<td>A.1</td>
<td></td>
</tr>
<tr>
<td>(Morton et.al. 1967)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Schork, 1964</td>
<td></td>
<td>2315</td>
<td></td>
<td>A.2,9,10</td>
<td></td>
</tr>
<tr>
<td>Cruz-coke and Covarrubias, 1964</td>
<td>Brazilian</td>
<td>-</td>
<td></td>
<td></td>
<td>C.1</td>
</tr>
<tr>
<td>Schull and Neel, 1965</td>
<td>Japanese</td>
<td>5984</td>
<td>A.1,2,4,6,</td>
<td>C.2,3,4,11,13</td>
<td>C.8,9,17</td>
</tr>
<tr>
<td>Neel et.al., 1970</td>
<td>Japanese</td>
<td>1082</td>
<td></td>
<td>A.1,4,6,7,8,12,28,C.2,4,11,12</td>
<td>C.1</td>
</tr>
<tr>
<td>Niswander and Chung, 1965</td>
<td></td>
<td>4740</td>
<td></td>
<td></td>
<td>A.13</td>
</tr>
<tr>
<td>Schreider, 1967</td>
<td>French</td>
<td>-</td>
<td></td>
<td>A.1</td>
<td></td>
</tr>
<tr>
<td>Krieger, 1969</td>
<td>Brazilian</td>
<td>3465</td>
<td></td>
<td>C.6</td>
<td>C.1</td>
</tr>
<tr>
<td>Neel et.al., 1970</td>
<td>Japanese</td>
<td>1082</td>
<td>A.1,4,6,7,8,11,28,C.2,4,11,12</td>
<td>C.1</td>
<td></td>
</tr>
<tr>
<td>Barbosa and Krieger, 1979</td>
<td>Brazilian</td>
<td>-</td>
<td></td>
<td>C.2</td>
<td></td>
</tr>
<tr>
<td>Afzal, 1988</td>
<td>Indian</td>
<td>2156</td>
<td></td>
<td>C.11,16</td>
<td></td>
</tr>
<tr>
<td>Badaruddoza, 1990</td>
<td>Indian</td>
<td>364</td>
<td>A.1,2,8,9,C.11,16</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Agrawal et.al., 1984</td>
<td>Indian</td>
<td>100</td>
<td>C.11</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Basu &amp; Jindal, 1987</td>
<td>Indian</td>
<td>680</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>