HEALTH EFFECTS OF CONSANGUINITY IN PONDICHERRY

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ABSTRACT

Of 1000 pregnant women studied, consanguinity was observed in 30.8%, with a higher frequency among women from rural areas, and among Hindus. In the consanguineous group, first cousin marriages were present in 47.4%, and uncle niece marriages in 23.4% of women. Coefficient of inbreeding was highest in Harijans (0.0258), followed by non-Brahmins (0.0220) and Brahmins (0.0204). Fertility was not influenced by consanguinity. There was a significantly higher rate of still births and infant mortality in consanguineous matings as compared with non-consanguineous. Total morbidity was higher in the consanguineous group as compared with the non-consanguineous ($p<0.01$), especially that due to neonatal infections and jaundice. There was no significant difference in the prevalence of congenital malformations, chromosomal and genetic disorders between the two groups, although the number of abnormal births in this group was small.

Key words: *Consanguinity, Morbidity, Mortality, Stillbirths, Neonatal deaths, Congenital malformations, Genetic disorders.*

It is well known that consanguinity increases the chance of the husband and wife carrying an indentical gene derived from a common ancestor. Children of such a marriage, therefore, are at greater risk of being homozygous for a harmful gene, and consequently suffer from autosomal recessive genetic disorders(1). Studies in India tracing the pedigrees of consanguineous marriages and estimating the coefficient of inbreeding have been summarized by Roy Choudhury(2). There is limited work available on the biological effects of inbreeding(3-8). The present investigation was undertaken to determine the prevalence and type of consanguinity in Pondicherry, and its effects on neonatal and infant morbidity and mortality.

Material and Methods

We studied 1000 consecutive pregnant women admitted for delivery in the maternity ward; in JIPMER hospital during 1978. A detailed history was obtained from the mother about her past obstetrical record, and antenatal period of the present pregnancy. Presentation and mode of delivery were ascertained, the newborn was examined immediately after birth and again after 24 hours, for assessment of maturity and presence of congenital malformations. All

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Received for publication July 31, 1991;
Accepted May 6, 1992

live born babies were followed up postnatally during their stay in the hospital (mostly 6-7 days) for immediate neonatal morbidity and mortality. Of the 1000 newborns, 360 were followed for a period of 3 months at monthly intervals, for morbidity.

Marriages were classified into four types: (*i*) spouses who were first-cousins (cross cousins)--a man marrying his mother's brother's daughter (MBD), or marrying his father's sister's daughter (FSD); (*ii*) spouses related closer than first cousins, mainly uncle-niece; (*iii*) spouses whose relationship was beyond first cousins were designated as others; (*iv*) spouses who were not related to each other before marriage were designated as non-consanguineous. The mean co-efficient of inbreeding for different religious groups and castes, as well as for total population, was calculated as per Sewell Wright's method of path co-efficient(1).

Results

1. *Prevalence of consanguinity:* Of the 1000 couples, consanguinity was found in 30.8% (*Table I*). It was significantly higher in the rural as compared to the urban population (35.6% *vs* 25.9%, $p<0.001$). *Table II* shows the Hindus had a higher frequency of consanguineous marriages than Christians or Muslims ($p<0.001$). Among the Hindus, highest frequency of consanguinity was observed in Harijans (37.5%), followed by non-Brahmins (30.8%) and least in Brahmin communities (27.7%). However, the difference in the frequency of consanguinity among Brahmins, non-Brahmins or Harijans was not statistically significant (*Table II*).

2. *Type of consanguinity:* Majority of consanguineous marriages were between first cousins (47.4%). Uncle-niece marriages were present in 23.4%, while 29.2% of the marriages were among those with more distant relationship (*Table III*). Uncle niece marriages were not practised by Christians and Muslims, but were common in all Hindu communities.

The commonest first cousin marriage was between a boy and his mother's brother's daughter. All the marriages were between cross-cousins. No parallel cousin marriages were observed in this study.

TABLE I—*Prevalence of Consanguinity by Residence and Religion*

	Consanguineous		Non-Consanguineous		Total
	No.	%	No.	%	
Residence					
Rural	179	35.6*	323	64.4	502
Urban	129	25.9	369	74.1	498
Religion					
Hindus	297	32.3**	624	67.7	921
Christians	9	14.1	55	85.9	64
Muslims	2	13.3	13	86.7	15
Total	308	30.8	692	69.2	1000

* Rural *vs* Urban $p<0.001$

**Hindus *vs* Christians $p=0.004$

TABLE II—*Co-efficient of Inbreeding by Caste and Religion*

	First cousin 1/16	Uncle niece 1/8	Others	All consan-guineous	Non-consan-guineous	F
Brahmins	5	3	2	10	26	0.0204
Non-Brahmins	94 (45.4)	51 (24.6)	62 (30.0)	207	463	0.0220
Harijans	39 (48.8)	19 (23.2)	22 (27.5)	80	135	0.0258
Christians	7	–	2	9	55	0.0074
Muslims	1	–	1	2	13	0.0052
Total	146 (47.4)	72 (23.4)	90 (29.2)	308 (100)	692	0.0199

Figures on parentheses indicate % of the consanguineous group.
F : Coefficient of inbreeding.

TABLE III—*Neonatal and Infant Deaths and Consanguinity*

	Consanguineous (n = 308)		Non-consanguineous (n = 692)	
	No.	Per 1000	No.	Per 1000
Total live births	767		1793	
Neonatal deaths*	49	63.8	87	48.5
Infant deaths** (0-1 yr)	75	97.8	107	59.7

Consanguineous *vs* non-consanguineous.
* Neonatal deaths p = 0.135, ** Infant mortality p = 0.0007.

3. *Co-efficient of inbreeding by caste and religion:* The mean co-efficient of inbreeding in the population was 0.0199. It was found to be highest in various Hindu communities, and lower in Christians and Muslims (*Table II*). However, due to the small number of subjects among Muslims and Christians, the statistical comparison is not very meaningful. Among the Hindus, the Harijans hand the highest co-efficient of inbreeding (0.0258), followed by non-Brahmins (0.0220) and Brahmins (0.0204). These differences were not statistically significant.

4. *Effect of consanguinity on mortality:* Neonatal and infant mortality (upto 1 year of age) for the whole group were higher in consanguineous matings, as compared with non-consanguineous matings (*Table III*). This was computed from the history of previous births, and their outcome. However, the difference was statistically significant only for infant mortality (97.8 *vs* 59.7, $p<0.05$).

5. *Effect of consanguinity on obstetrical complications:* Obstetrical complications like pre-eclamptic toxemia (PET), anemia, Rh incompatibility, and antepartum hemorrhage were more among the consanguineous (47/308 = 15.2%), as compared with non-consanguineous matings (89/692 = 12.8%). However, the difference was not statistically significant (p = 0.36). The mode of delivery was not affected by consanguinity.

6. *Effect of consanguinity on fertility:* The mean total fertility rate per couple was 2.8 in both consanguineous and non-consanguineous marriages, showing that consanguinity did not affect fertility.

7. *Consanguinity and fetal loss:* Fetal losses which occurred at or before 28 weeks of gestation were 5.7 and 4.4 per 100 pregnancies, respectively, in consanguineous and non-consanguineous groups. Still births were also higher in the former than in the latter (4.2 *vs* 2.8/100 pregnancies, $p<0.01$). The data on fetal losses and still births was obtained from the obstetrical histories of the mothers.

8. *Effect of consanguinity on the baby:* Sex of the baby was not influenced by consanguinity. Mean weight of the newborns was also not affected by consanguinity. There was no difference in the frequency of asphyxiated babies between consanguineous and non-consanguineous group.

9. *Effect of consanguinity on neonatal morbidity (Table IV)*: (*a*) The *frequency of illnesses* was significantly higher in offsprings of consanguineous marriages as compared to non-consanguineous marriages (176.1/1000 *vs* 67.2/1000, $p<0.001$). Consanguinity significantly increased the prevalence of severe neonatal infections and septicemia ($p<0.001$). Jaundice was also observed in a significantly greater number of consanguineous newborns (106.3 *vs* 67.2 per 1000, $p<0.001$).

(*b*) *Congenital malformations:* Consanguineous marriages showed a higher frequency of malformations in the offspring, when compared to the non-consanguineous marriages. However, this did not reach statistical significance (p = 0.3). The various malformations that were observed were: hydrocephalus (n = 3), tracheo-esophageal fistula (n = 1), hydrocele (n = 1), epispadias (n = 1) it in the consanguineous group, and congenital talipes equinovarus (n = 3), cleft lip (n = 1), hydrocele (n = 1), congential bladder neck obstruction (n =1), chordee penis (n = 1), congenital cyanotic heart disease (n = 2) in the non-consanguineous group.

(*c*) Three *chromosomal anomalies* were observed in the non-consanguineous group, while no such abnormality was seen in the consanguineous group.

(*d*) *Autosomal recessive disorders:* One case of albinism was observed in the consanguineous group, while congenital adrenal hyperplasia was present in a baby in the non-consanguineous group.

10. *Morbidity on follow-up (n = 360):* The number of illnesses recorded were significantly greater in the consanguineous offspring, as compared with the non-consanguineous. The number of illnesses like-respiratory infections, skin infections, gastro-intestinal infections, *etc.* were higher in offsprings of consanguineous marriages (60/111 = 54.1%) as compared to non-consanguineous couples (59/249 = 23.7%, $p<0.001$).

Discussion

Inbreeding has a higher prevalence in South India, especially in Tamil Nadu and Pondicherry. The frequency of consanguin-

TABLE IV—*Effect of Consanguinity on Morbidity in Early Neonatal Period*

Types of illnesses	Consanguineous		Non-consanguineous	
	No.	Per 1000 live births	No.	Per 1000 live births
Infections and septicemia	16	53.2*	8	11.6
Acute suppurative otitis media	–	–	2	2.9
Birth injuries	2	6.6	9	13.1
Aspiration syndromes	2	6.6	7	10.2
Respiratory distress syndrome	1	3.3	–	–
Neonatal jaundice	32	106.3**	20	29.2
	53	176.1***	46	67.2
Genetic disorders				
Autosomal recessive disorders	1	3.3	1	1.45
Chromosomal anomalies	–	–	3	4.4
Congenital malformations	6	19.9****	9	13.1
	7	23.2	13	18.97

Consanguineous *vs* non-consanguineous:
*Infections—$p < 0.001$, ** Jaundice—$p < 0.001$, *** All illnesses—$p = < 0.001$,
***** Malformations—Not significant.

ity in the present study was 30.8%, which is less than that observed in other studies in South India(3,5-7). This lower frequency could be due to the fact that it was a hospital-based study. The higher prevalence in the rural group is consistent with the data from other studies(9,10). Hindus showed a higher frequency of consanguinity as compared with Christians and Muslims, which can be explained by the strong cultural and traditional factors operating among the Hindus. Though consanguineous marriages are prohibited by law of Church among Roman Catholics, consanguineous marriages do occur among them; perhaps because most of them are converts from Hindus and they still follow certain older traditions. Illiteracy, ignorance and rural influence probably explain the high frequency of consanguinity found among Harijans.

The most frequent type of consanguineous marriage in our study was between first cousins (47.4%), especially that between a boy and his mother's brother's daughter. The frequency of first cousin marriage varies from 5 to 57% in different parts of South India depending upon the place of study(2,7,11-14). All the cousin marriages were between cross cousins. No parallel cousin marriages were observed. Uncle-niece marriages were observed only among Hindus. These are not seen in other religious groups in India(2).

The degree of consanguinity to which an offspring is inbred is measured by the co-efficient of inbreeding (F). It is defined as the probability that two genes in one offspring are identical by virtue of their descent from one common ancestor. The mean co-efficient of inbreeding was highest

in Harijans (0.0258), followed closely by non-Brahmins (0.0220) and Brahmins (0.0204). It was low in Christians and Muslims. These findings are in agreement with other studies(2,7,12,15).

In the present study, consanguinity did not affect total fertility, although rate of stillbirths and infant mortality in the 0-1 year age group was significantly increased. Fetal losses were higher in the consanguineous group, although as compared with non-consanguineous the difference did not reach statistical significance. Generally studies in India show that gross fertility is increased in the consanguineous groups(7,16,17), although Rao in Vellore did not find any significant increase(5). The increased fertility is partly due to early age at marriage, and a longer reproductive lifespan(7,17).

Most studies have demonstrated higher fetal losses, neonatal deaths and infant mortality among pregnancies and their outcome in women who are married consanguineously(17,18). Three studies on newborns have reported increased total mortality (abortions, stillbirths and infant mortality(19-21). In the study by Stevenson *et al.*(22) too the data from Calcutta and Bombay documented a higher frequency of stillbirths and neonatal deaths in the offsprings of consanguineous marriages. Contrary to these, Rao from Vellore(5,23) did not observe any significant influence of inbreeding on fetal losses and neonatal deaths, although infant mortality was increased. Bittles *et al.*(18) also demonstrated a greater post-neonatal mortality rate in Bangalore based on retrospective analysis.

In the present study neonatal morbidity was significantly increased in the consanguineous group. This was especially evident for severe infections and septicemia, and pathological jaundice. The frequency of illnesses in the first 3 months of life was also increased among the consanguineous infants.

In the present study, congenital malformations were marginally higher in the consanguineous offspring as compared with the non-consanguineous, although statistical significance was not achieved possibly due to small number of cases. The Indian data, on the frequency of congenital malformations, summarized elsewhere(24), showed a one and a half times higher frequency among consanguineous matings as compared with the non-consanguineous. Kulkarni and colleagues(25) also demonstrated a higher rate of neural tube defects among the consanguineous families in Karnataka, supporting the earlier observations from the study of Stevenson *et al.*(22) in Egypt and elsewhere.

The sample size in the present study was not sufficiently large to provide enough infants being born with chromosomal and genetic disorders among the consanguineous and non-consanguineous groups. However, Puri *et al* in an earlier hospital-based study(26) showed that frequency of Down syndrome was higher amongst offsprings of consanguineous marriages. Similarly, in another population based study Puri and colleagues(17) reported a higher prevalence of autosomal recessive disorders. However, Sanghvi(4,12) suggested that in South Indian communities wherein consanguineous marriages have been practised over many generations, there has been a weeding out of deleterious autosomal recessive genes. Therefore, many population based studies do not show any clinically significant effect of consanguinity(23,27). For example, Appaji Rao and colleagues(27) who screened over 100,000 newborns for the presence of aminoacid

disorders did not find an increase in consanguinity in those who had aminoacid disorders. Our experience with aminoacid screening of children with mental retardation at Delhi (Kaur and Verma, unpublished observations) showed that in three recently diagnosed cases of maple syrup urine disease and one case of citrullinemia there was no consangunity among the parents. Therefore, this cannot be used as an argument that consanguinity is not harmful. It only means that the frequency of the genes for these disorders is fairly common in the general population, so that marriage between two carriers occurs even in a random mating.

It may be concluded that in India consanguinity is likely to be one of the contributory factors for higher infant morbidity and mortality. It would be advisable to avoid consanguineous marriages in families, where already a child with autosomal recessive disorder has been born and to reduce consanguineous marriages in the others as far as possible.

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