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Effect of Consanguinity on the Outcome of Pregnancy

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INTRODUCTION

Inbreeding in South India has been attributed to the inherited Dravidian culture (Jacob and Jayabal, 1971). Reviews on the effects of inbreeding on morbidity and mortality resulting in detrimental reproductive outcome are plenty. During internship, the authors carried out this study as part of the 3 months rural posting in the villages, in the outskirts of Bangalore. Prevalence of consanguineous marriages in the community around the rural center and the effect of consanguinity on the pregnancy outcome have been studied, keeping in mind factors such as socio-economic status and education.

MATERIAL AND METHODS

All together, 150 families have been interviewed. The degree of consanguinity was described by the inbreeding coefficient that is equal to the probability that the two genes, which the individual has at a given locus, are identical by descent (Jacob and Jayabal, 1971) (Table 1).

Table 1: Inbreeding coefficient

Description	*Inbreeding coefficient*
Uncle niece marriage	1/8
First cousins	1/16
First degree step cousins	1/32
First cousins once removed	1/32
Second cousins	1/32

Hence in our study uncle-niece and 1st cousin marriage have been considered as consanguineous and 2nd and 3rd cousins unrelated. 1st cousins were further divided into FSD (Father's sister's daughter) and MBD (Mother's brother's daughter). The outcomes of pregnancy have been analysed. Socio-economic status of these families was found to be similar. A detailed proforma covered the name, age, address, duration of married life, pregnancy outcome, minimum 3-generation pedigree and consanguinity. Mother was personally and extensively interviewed and pregnancy history including livebirth, abortion (foetal death-before 28 weeks of gestation), stillbirth (foetal death after 28 weeks), neonatal death (early/late), post neonatal death, (upto 5 years of age), specific and non-specific causes of these deaths and any malformations/deformities in dead or live born has been obtained.

Association betwen the parental consanguinity and the pregnancy outcome was analysed, with chi-square test.

RESULTS

The frequency and the type of consanguinity is shown in table 2. Out of 150 families 89 (59.33%) were non-consanguineous and 61 (40.67%) consanguineous. In consanguineous group 47.54% were Uncle - Niece (U-N) (29) and 52.46% (32) 1st cousins.

High rates of abortions and IUDs (Intra uterine deaths)were found among the pregnancy in MBD group (12.3% and 6.7%) as compared to that of FSD group (2.5% and 2.5%). (Table 3)

Table 2: Frequency and types of consanguinity

Type	*Type of consanguinity*	*Number*	*%*	
Consanguineous	U-N	29	19.33%	61(40.67%)
	I cousin	32	21.33%	
Unrelated	II cousin	2	1.33%	89(50.33%)
	III cousin	3	2.0%	
	Non-Consanguineous	64	56.0%	

Table 3: Effect on pregnancy outcome depending on type of first cousins

Type	*Family 1*	*Pregnancies*	*Livebirths*	*Abortions*	*IUD*	*Deformities*
FSD	14(43%)	339	33	1(2.56%)	1(2.56%)	1(2.56%)
MBD	18(57%)	65	51	8(12.3%)	4(6.15%)	0
Total cousin	32	104	84	9	5	1

FSD-Father's sister's daughter MBD-Mother's brother's daughter

Table 4: Effect on maternal age at first pregnancy on pregnancy outcome

Age group	*Family 1*	*Pregnancies*	*Livebirths*	*Abortions*	*IUD*	*Deformities*
12-17	75(50%)	281	247	15(5.3%)	7(2.4%)	7(2.4%)
18.26	75(505)	220	191	17(7.7%)	2(0.9%)	5(2.7%)

FSD-Father's sister's daughter MBD-Mother's brother's daughter

Table 5: Measure of fertility in consanguineous and unrelated marriage

		Consanguineous			*Unrelated*
Particulars		*Uncle-Niece*	*Cousin*	*Total*	
Families	Total	29(47.54%)	32(52.46%)	61(40.67%)	89(59.33%)
Pregnancy	Total	104(50%)	104(50%)	208(41.52%)	293(58.48%)
	Normal	77(74.04%)	80(76.92%)	171(82.21%)	257(87.71%)
Live birth	Total	88(84.61%)	84(80.76%)	172(82.7%)	267(91.13%)
	Normal	79(89.77%)	83(98.81%)	162(94.18%)	265(99.25%)
Surviving offspring		81	75	156	216

In our study, the lowest maternal age at pregnancy was 12 years and the highest was 26 years. Maternal age was grouped into less than 18 and more than 18 years of age. It was found that the maternal age at the first prenancy had no significant effect on the pregnancy outcome (Table 4)

In Table 5 is shown the measures of fertility. In a total of 150 families, 501 pregnancies were recorded. The number being 208 (41.32%) in consanguineous and 293(58.48%) in the unrelated group. Total number of live born was 439, that of normal live born (i.e. without any obvious deformities) was 427. 94.18% (162) in the consanguineous group and 99.25% (265) in the unrelated group. The number of surviving offspring (total live born minus non-accidental deaths before the age of 21 years) was 156/172 and 216/267 in the consanguineous and in the unrelated group respectively.

Analysis of reproductive wastage in table 6 showed increased abortions 8.65% and still birth 3.85% in the inbred as compared to 4.78% and 0.34% respectively in the unrelated group ($p<0.05$ for abortion; $p<0.001$ for still birth). The frequency was more in the U-N marriages.

Infant mortality (neonatal and post neonatal deaths) and child mortality were observed to be more in the unrelated group. However this was not statiscally significant (Table 7).

Congenital malformation was found in 4.8% of consanguineous and 0.68% in unrelated group ($p<0.001$). It was higher in the U-N(9/501) than in the 1st cousin (1/501) (Table 8). (spina bifida, mental retardation, Down syndrome, cardiovascular

Table 6: Pregnancy wastage according to the degree of parental consanguinity

Type	*Consanguineous*			*Unrelated*	*'p' value*
	Uncle-Niece	*1st Cousin*	*Total*		
No. of Pregnancies	104	104	208	293	
Abortions	9	9	18(8.65%)	14(4.78%)	<0.05*
Still Births	3	5	8(3.85%)	1(0.34%)	<0.001*
Total Wastage	12	14	26(12.5%)	15(5.12%)	<0.001

* Significant

Table 7: Infant and child deaths according to the degree of parental consanguinity

Type	*Consanguineous*			*Unrelated*
	Uncle-Niece	*1st Cousin*	*Total*	
No of Pregnancies	104	104	208	293
No. of live births	88	84	172	267
Neonatal Deaths	3	7	10(5.81%)	16(5.99%)
Post Neonatal Deaths	1	1	2(1.616%)	5(1.87%)
Total Infant Deaths	4	8	12(6.98%)	21(7.86%)
1-4 yr Mortality	3	1	4	8

Table 7: Live births-congenital malformations

	Consanguineous			*Unrelated*	
	Uncle-Niece	*Cousin*	*Total*		*'p' value*
Spina bifida	2	0	2	0	
CNS	4	0	4	0	
CVS	2	0	2	0	
Musculo skeletal	1	1	2	0	
Cleft-lip	0	0	0	1	
Cong deaf muslim	0	0	0	1	
Total	9	1	10(4.8%)	2(0.68%)	<0.001*

* Significant

malformation such as congenital heart diseases, musculo-skeletal including club foot and Radio-ulnar aplasia, cleft lip and congenital deaf Muslim).

DISCUSSION

Reviews on the effects of consanguinity on the reproductive outcome, fertility, mortality, congenital malformations and fetal growth/development/health have been published. Published reports are conflicting since such effects may be determined by genetic as well as environmental factors. It may be difficult to specifically identify the genetic disorders and to determine their incidence against the prevailing background of infection and nutritional disease.

In this study, among the types of consanguinity first cousin relationship was found to be prevalent as reported by Bittles et al., 1991; Verma et al., 1992 Al-Gazali et al., 1997.

A higher rate of fertility was observed in the consanguineous group. It could be due to the younger ages at marriage; which may have increased the reproductive life span of the couples, especially that of the women (Indra et al., 1977; Basu, 1978; Hussain and Bittles, 1999). The overall picture of younger maternal age, at first pregnancy, with greater fertility is in line with the studies by Basu (1978), Bittles et al. (1991); Indra Bai et al. (1997), and Hussain and Bittles (1999).

In this study, an increased incidence of abortions ($p<0.05$) and still births ($p<0.001$) have been found in the consanguineous group. In fact, in the first cousin families, still births were found to be more. These observations are consistent with other studies in literature by Jacob et al. (1971), Hussain and Bittles (1991), Verma et al. (1992), Yasim et al. (1997), Stoltenberg (1999). However Shivkumaran et al. (1997), Demirel (1997) and others have not found any statistical significance (Demirel et al., 1997; Jaber et al., 1997; Sivakumaran and; Al Abdul kareem and Ballal, 1998).

Effect of consanguinity on the infant and child mortality was found to be significant by Yadsim et al. (1997), Jaber et al. (1997) and Yaqoob et al. (199). However in the present study it was not significant.

Incidence of congenital malformations was found to be more in the consanguineous group ($p<0.001$) and that too in the uncle-niece families. Similar observation has been made by Maderi et al. (1979), Karimi-Nejad et al. (1991) Demirel et al. (1997) and Martinez-Friyaz et al. (1998). This could be explained on the basis of the inbreeding coefficient where the risk is 1/8 for U-N when compared to 1/16 for 1st cousin (Murthy and Jamil, 1972).

The non observance of malformation in non consanguineous group may indicate that there could be a beneficial effect in the reduction in the number of deleterious gene (Sanghvi, 1982). It may appear better to dissuade the prospective marriages in consanguinity. An attempt to break up the marriage with long tradition should not be initiated until and unless all possible social consequences have been carefully considered and medical / genetic problems have been eliminated.

KEY WORDS Consanguinity. Pregnancy outcome. Congenital Malformations.

ABSTRACT In this study the frequency of the consanguinity and its effects on the pregnancy outcome in a selected study population has been attempted. A total of 150 families consisting of Kannadigas (62.3%), Tamilians (23.21%), Telugus (13.24%) and Malayalis (1.25%) were interviewed. Their economic status was found to be more or less similar. Out of which, 40.67% (61) were consanguineous and 59.33% non-

consanguineous (89). In consanguinity. 1st cousin relationship was observed in 32(52.46%) and 29 were uncle-niece (47.54%). Out of 32.1st cousins 43% were FSD (father's sister's daughter) and 57% were MBD (mother's brother's daughter) Pregnancy wastage was found to be higher in the consanguineous group ($p<0.001$). The number of offspring with congenital malformations was also higher in the consanguineous group ($p<0.001$). Significant difference has not been observed in the occurrence of infant mortality.

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