RESEARCH ARTICLE

CONSANGUINITY STUDY IN HOSPITAL POPULATIONS OF PALAKKAD DISTRICT, KERALA I. PATTERNS, PREVALENCE AND EFFECTS

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SUMMARY

Consanguinity study was carried out involving the patients of three hospitals in the Palakkad district, Kerala. Patients belonging to only three groups (Nair, Chetty, Scheduled Castes and Scheduled Tribes) comprised numbers amenable for analysis. Aspects such as consanguinity patterns, frequency, effect of consanguinity on couple fertility, pre-reproductive mortality and morbidity in the three groups were assessed and evaluated. The most predominant pattern in all the three groups was first cousin type, with decisive preference for matri-lateral cross subtype. Frequency of consanguinity was relatively low in the Nair (10.04%) and medium in the Chetty (26.95%) and SC-ST (22.07%). Differentials of couple fertility were moderate and not significant. The mortality effect of consanguinity was fairly high and highly significant at all levels, suggestive of notable harmful effects of the phenomenon. The morbidity frequency was low, and the differential significant only in the Chetty group. Morbid conditions, particularly cleft lip-palate defect in the consanguineous Chetty group is striking and suggestive of genetic predisposition with potential association with consanguinity.

Keywords: Consanguinity, hospital populations, Palakkad district, effects.

INTRODUCTION

Consanguinity or human inbreeding is the phenomenon of marital union between spouses who are related to each other by common ancestry. The detrimental effects of blood-related marriages arise as a consequence of genetic homozygosis of deleterious recessive genes concealed in heterozygous state, which find expression as affected, homozygous offspring in higher frequencies in consanguineous families (Bittles & Hussain 2000). The affected offspring are likely to meet with developmental hazards and abnormalities *in utero* or after birth, and become nonviable leading to mortality at various pre-reproductive stages, and morbid conditions of genetic predisposition (Mathew 2017). The magnitude of consanguinity-related risk effects are dependent primarily on the nature, number and frequency of deleterious recessive genes present in the gene pool of populations, and on the degree, level and duration

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of inbreeding (Mathew & Jyothilekshmi 2017a) which are often influenced by a vista of socio-economic correlates and demographic determinants (Mathew 2018). A quantum of data exists globally highlighting the consanguinity-related risk effects in diverse population groups. Many castes and communities and tribals of the State of Kerala have been known as practising close-kin marriage alliances as part of their social custom and tradition for centuries. Extensive consanguinity studies carried out in Kerala communities have yielded a rich store of data suggestive of the social, fertility and genetic effects of the phenomenon (Mathew & Jyothilekshmi 2017a).

The consanguinity data and the effects thereof are conventionally assembled community-wise by survey method of data collection. However, several studies are known globally, involving inpatients of hospitals in Saudi Arabia (Shafi et al. 2003). Quatar (Bener et al. 2009), UK (Kilarski et al. 2012) and India (Attumalil et al. 2011, Bellad et al. 2012). The present paper concerns consanguinity study involving the patients of three hospitals in the Palakkad district of Kerala, aimed at assessing the patterns, frequency and effects of consanguinity based on data from the patients of three groups (Nair, Chetty and Scheduled Castes and Scheduled Tribes) which abounded in number amenable for analysis.

MATERIALS AND METHODS

Consanguinity data were collected from the patients of three government hospitals in the Palakkad district such as the District Hospital of Palakkad, the Government Hospital for Women and Children and the Community Health Centre at Nemmara. The District Hospital is one of the multi-specialty hospitals in the district, located at Sultanpet, with 544 beds. The government hospital for Women and Children, with 250 beds, is located near the District hospital, Palakkad. Nemmara is a rural area, which is the downtown of Nelliampathy Hill Station. The people of Nemmara and Nelliampathy, who are mainly low-income group farmers and labourers, depend on the Community Health Centre for their hospital service. The Health Centre is provided with 93 beds and five doctors. Consanguinity data were collected using a comprehensive questionnaire which includes a variety of socio-economic, demographic, reproductive and genetic parameters. The data of adult patients were collected directly from them, and in the case of children, from their bystanders, who are either their own parents or close relatives. Marriages contracted between spouses who are related in different degrees such as Uncle-Niece (U-N), First Cousin (1C), First cousin once removed (1.5C), Double first cousin (DFC) and Second cousin (2C) were classified as consanguineous, and those beyond second cousin as non-consanguineous. The first cousin pattern is further subdivided by a second level such as Patri-lateral parallel cousin (PPC), Matri-lateral parallel cousin (MPC), Patri-lateral cross cousin (PCC) and Matri-lateral cross cousin (MCC). The consanguinity factor was measured in terms of the frequency, Coefficient of consanguinity (F) and mean F which is calculated after Wright (1922).

Pre-reproductive mortality was considered under two classes, the prenatal (abortion, miscarriage, still birth) and postnatal (neonatal, post-neonatal, infant, juvenile). Total mortality is the sum of losses from abortion to juvenile mortality. Morbidity includes various congenital defects (physical, sensory, mental, systemic) and diseases. As the data of individual morbid conditions were too small, only the pooled data of all the conditions taken together considered. Couple fertility was measured in terms of three parameters such as conception, live-birth and child survival, and the mean fertility computed and assessed. The data from the patients were taken by periodical visits to the three hospitals at regular intervals of one month for one year during 2016-17. Data collected constitute a total of 2316 patients belonging to diverse social classes (Forward, Backward, SC-ST). The patients constitute a cross section of various communal groups of the district. As the number of patients of most of the communities was too small, only three groups with sufficient number amenable for analysis such as Nair (966), Chetty (616)

and SC-ST (734) were considered for a detailed study. Test for proportion was used to find out the significance of the effect of consanguinity, and the test for equality of means employed to test the significance of the difference in fertility rate and survival rate between the test and control groups.

OBSERVATIONS

The consanguinity patterns and their frequencies in the families of patients of the Nair, Chetty and SC-ST groups are furnished in Table 1, and the consanguinity effects on couple fertility, pre-reproductive mortality and morbidity in Table 2. The common consanguinity patterns were First cousin (1C) First cousin once removed (1.5C) and Second cousin (2C), of which the 1C type most prevalent, in which the matrilateral cross subtype (MCC) the most preferred. The frequency was low in the Nair and medium in the other two groups. Couple fertility profiles were moderate and differentials marginal. The total mortality risk effect was fairly high with highly significant differentials. The pooled frequency of morbidity was low. Incidence of cleft lip-palate defect in the consanguineous families of the Chetty group was striking.

TABLE 1: The patterns, frequency and mean F of consanguinity in the Nair, Chetty and SC-ST patients of Palakkad hospitals.

Pattern	Frequency of consanguinity (%)		
	Nair	Chetty	SC-ST
U-N	0	5.03	2.59
PPC	0.1	0	0.54
PCC	2.17	5.03	4.63
MPC	0.52	0	1.5
MCC	3.52	7.14	5.45
I C TOTAL	6.31	12.17	12.12
1.5 C	1.14	2.76	2.86
2 C	2.59	6.98	4.49
C	10.04	26.95	22.07
NC	89.96	73.05	77.93
Mean (F)	0.005 ± 0.001	0.0159 ± 0.0013	0.0124 ± 0.001

C - Consanguineous, NC- Non-consanguineous.

TABLE 2. Effect of consanguinity on mortality, morbidity and couple fertility in the families of patients in the Palakkad hospitals.

Community	C%		Mortality %			Mb %	Mean fertility
			Prenatal	Postnatal	Total		
Nair	10.04	C	11.2 [**]	3.45 [*]	14.65 [**]	5.56 [ns]	2.39 [ns]
		NC	3.63	0.66	4.29	2.93	2.09
Chetty	26.95	C	12.95 [**]	3.57 [**]	16.52 [**]	8.29 [**]	2.70 [ns]
		NC	2.59	0.83	3.42	1.44	2.4
SC-ST	22.07	C	12.26 [**]	3.4 [**]	15.66 [**]	6.93 [ns]	3.27 [ns]
		NC	3.77	0.99	4.77	4.24	2.64

C - Consanguineous, NC- Non-consanguineous, Mb- Morbidity. * Significant at 0.05 level, ** Significant at 0.01 level, ns - Not significant .

DISCUSSION

The common patterns of consanguinity known in diverse population groups the world over, in the order of spousal relationship are, Uncle-Niece (UN), First cousin (1C), First cousin once removed (1.5C) and Second cousin (2C), of which the 1C type is the most preferred in all. Although the four sub-types of first cousin pattern have the same genetic relationship, discrimination exists based on specific customs. Except the Muslims of the Middle East who prefer Patrilateral parallel cousin (PPC) marriages, all the others the world over prefer the cross cousin types, especially the Matrilateral cross (MCC) pattern. In all the Indian communities, including the innumerable ones belonging to diverse social classes and tribals studied earlier from the State of Kerala (Mathew et al. 2006, Mathew & Jyothilekshmi 2017a, b), the same practice prevails, and this is so with all the three groups presently studied from the Palakkad hospitals (Table 1). Here the closer degree Uncle-Niece (UN) is absent in the Nair group, but present in the other two in appreciable frequency. The level of inbreeding is measured in terms of two parameters, the frequency and the coefficient of inbreeding (F). The former is the rate at which the different patterns together occur, calculated as percentage of the total sample, while the latter is defined as the probability that the consanguineous offspring is an identical homozygote. The coefficient values of the different patterns in the Palakkad hospital groups are 1/8 for Uncle-Niece, 1/16 for First cousin, 1/32 for First cousin once removed and 1/64 for second cousin. The frequency of consanguinity in the present three groups are low (10.04%) in the Nair and medium in the other two such as 26.95% (Chetty) and 22.07% (SC-ST). A wide range of rate levels exists in the world scenario, ranging from less than 0.5% to as high as 97%, the lowest occurring in USA and in most European Christian countries in which the low rates are attributable to the impact of Church ban on closer degree alliances up to 2C, coupled with increased mobility, urbanization and consequent socio-economic development (Bittles 2012). The frequencies in the Kerala group of communities earlier studied range from 1.55% in the Christian Mala Arayas to a very high of over 90% in certain tribes in the Idukki district, of which the highest ever reported (97%) occurs in the Kurumba Pulaya tribe (Mathew & Jyothilekshmi 2017a, Sindu et al. 2001). The consanguinity rates and mean Fs in the Kerala communities have revealed a clear and consistent gradient, very much associated with socio-economic co-variables and the literacy factor. This appears to be very much so with the present three groups in which the socio-economically and educationally more advanced group (Nair) has registered the lowest frequency (10.04%) and mean F (0.005 ± 0.001).

The overall fertility data in the Kerala communities earlier studied showed significant differentials between the consanguineous and non-consanguineous families (Mathew & Jyothilekshmi 2017a). Available information on the association between consanguinity and fertility profiles has projected varying trends, in which the socio-economically developed communities registered very low fertility means, with only marginal differentials (Mathew et al. 2006). The fertility means evident in all the present groups were low and the differentials not significant and marginal (Table 2). Studies in different population groups have projected conflicting suggestions and divergent conclusions regarding the consanguinity-associated mortality effects, some indicating notable harmful effects, others no effect, and still others even beneficial effects. In the present groups, all registering highly significant

differentials, the mortality effect by inbreeding is clearly harmful. Prenatal mortality refers to pregnancy wastage by foetal loss at various stages before birth. It has been estimated that a sizable proportion of human conceptions are lost before implantation or shortly afterwards (Robert & Ian 1998). The present data in all three groups show highly significant prenatal differentials. Several factors are known to contribute to high rates of foetal loss in consanguineous families, which include increase of homozygosity of lethal recessive genes that affect growth and development of the embryo. Post-natal mortality refers to loss of offspring at different stages after birth. Post-natal differentials in two of the present groups (Chetty, SC-ST) are highly significant, while less significant in Nair group. The pre- and post-natal mortality data in Kerala communities studied earlier have displayed two trends, either pre-natal in excess or vice versa (Mathew et al. 2006). In the present data, the pre-natal is in excess consistently in all the three groups. Various reasons have been proposed as explanation for the pre-natal excess, which include genes responsible for fetal loss are higher in number. The total mortality rates in the present groups range from 14.65% (Nair) to 16.52% in the Chetty group, and the differentials in all the three groups are highly significant which is suggestive of notable consanguinity effect on mortality. Morbidity refers to various kinds of diseases and congenital defects/ abnormalities involving different organs or organ systems, constituting a variety of detrimental conditions. The morbid conditions noticed in the patients and family members of present study are many, which can be classified as physical (dwarfism, microcephaly, hydrocephaly, cleft-foot, cleft lip/ palate), sensory (deaf, dumb, squint eye, blindness), mental (mental retardation, epilepsy, schizophrenia, Alzheimer's) and systemic (tuberculosis, diabetes, cancer, cardiac, bronchitis etc). The pooled morbidity data in the present three groups (Table 2) showed relatively low rates ranging from 5.56% to 8.29%, the differentials being highly significant only in the Chetty group. A number of consanguinity studies are known globally based on hospital data, of which many from the Middle East and a few from India, including Kerala; and in many studies, specific human defects and diseases were implicated to consanguinity: eg, Cleft lip-palate from Saudi Arabia (Shafi et al. 2003), Turkey (Altunhan et al. 2012), cancer from Qatar (Bener et al. 2009), Parkinson's disease from UK (Kilarski et al. 2012), metachromatic leucodystrophy in children from Bengaluru, India (Bindu et al. 2005), and childhood epilepsy from Thiruvananthapuram, Kerala (Attumalil et al. 2011) etc. The data from the present Women and Children hospital which showed seven cases of children with cleft lip-palate defect of which five from the consanguineous families of the Chetty group appears to signify clear association of the defect with consanguinity. Bittles (2012) who has assembled massive morbidity data from hospital populations globally, held that in most cases the defects/diseases are genetically predisposed with potential association with inbreeding.

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