Impact of consanguinity on cardio-metabolic health and other diseases: findings from an Afro-Indian tribal community

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Abstract Numerous anthropological reports have indicated consanguineous marriage in populous Asian countries, but the overall impact of first cousin marriage on survival and health of specific communities has rarely been reported. The aim of the study was to estimate risks for various complex diseases in the progeny of consanguineous parents. A cross-sectional study was conducted among 222 women among Siddis, a particularly vulnerable tribal group in the state of Gujarat, India, who are Sunni Muslims by faith. The Siddis are not part of the original Negrito element of India. They are descendants of Africans from Northeast and East Africa who were brought to India as slaves, soldiers, or servants. The degree of consanguinity between each female and her spouse and the degree of consanguinity between their parents and proband's grandparents were recorded with the help of pedigrees. The results showed that the rate of consanguinity in the present generation was 49 % ($N=109$), higher than preceding generations. A significant association was observed between women's age, educational level, occupational status, consanguineous parents, and consanguinity. Socioeconomic status and consanguinity showed U-shaped associations. Nearly three times odds for cardio-metabolic risks (2.65 odds ratio (OR) for heart diseases, 2.44 OR for diabetes mellitus, and 2.62 OR for hypertension) have been contracted in the progeny of consanguineous marriage in the parental generation. The risk of cardio-metabolic diseases is higher in offspring of consanguineous couples, and there is a significant increase in the prevalence of common adult diseases.

Keywords Consanguinity · Afro-Indians · Epidemiology · Cardio-metabolic health

Abbreviations

SLI Standard of living index

Introduction

Consanguinity refers to a union between two people who share an ancestor or share blood. The term consanguineous is derived from the two Latin words “con” meaning shared and “sanguis” meaning blood. Globally speaking, one fifth of the human population around the world lives in communities with a preference for consanguineous marriage, and at least 8.5 % of children have consanguineous parents (Modell and Darr 2002). In many regions of Asia and Africa, consanguineous unions currently account for approximately 20 to 50 % of all unions (Bittles et al. 1991), yet limited quantitative information is available on consanguinity. The data is even scarcer when interpreted in terms of caste, ethnicities, and religion from particular regions, especially India.

The demographic population in India has been divided into innumerable endogamous units, linguistic groups, religious sects, occupations, local divisions, and sub-divisions within communities (Mukherjee 1973). Each of these groups has its own marriage pattern based on survival possibilities, preferential rules, religious prescriptions, and socioeconomic circulation ethos. Similarly, the Muslim population of India is subdivided into Sunni, Shia, Ismaili and Dawoodi Bohra communities, and biraderis that are based on traditional social and occupational divisions. Consanguineous marriage is common in all Indian Muslim communities (Bittles and Hussain 2000).

Consanguinity is seen to have copious correlates, and this type of marriage has been a long-standing habit among

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various communities in India. While the level of inbreeding depends on several factors including demographic, religious, cultural, environmental, and socioeconomic factors, the roots of these unions lie in reasons like strengthening of family relationships, with economic benefits an additional consideration; the small size of the local population; advanced bridal age; or lack of dowry (Cavalli-Sforza et al. 2004; Bittles 2001; Bittles 2002).

Numerous anthropological reports have indicated consanguineous marriage throughout Sub-Saharan Africa and in populous Asian countries, but the overall impact of first cousin marriage on survival and health on specific communities has been rarely reported (Bittles and Black 2010). Contemporary attention on consanguineous unions is largely focused on the expression and identification of rare autosomal recessive alleles and their impact on mortality and fertility. An increased homozygosity can lead to an increased risk of premature morbidity and mortality among the offspring. There is a dearth of data on associations between consanguinity and cardio-metabolic diseases.

Also, it is a challenging task to find an endogamous, homozygous group practicing consanguinity. Siddis are enlisted as a “particularly vulnerable tribal group” in the Indian constitution. There has been a deficiency of comprehensive health research among the tribal populations of India especially in terms of affinal associations and the consequences. There seems an urgent need for initiating area-specific, tribe-specific, and action-oriented health research in consonance with the felt needs of the tribal communities as they are experiencing an increase in cardio-metabolic disorders and associated health problems. The research might help in the construction of life course models for prevention strategies with the type of marriage being one unexplored pathway. Hence, the present study was undertaken to estimate risks for various complex diseases in the progeny of consanguineous parents.

Methods

Target population

A cross-sectional study was conducted among the Siddis, a particularly vulnerable tribal group in the state of Gujarat, India, in October, 2011, to determine the impact of consanguineous marriages on cardio-metabolic health and other common diseases. A multistage stratified cluster sampling design was developed using district census of Junagadh district of Gujarat, India. Five reasonably populated villages (population ≥ 1000), namely Jambur, Sirvan, Javantri, Surva, and Talala (Talala Taluka), Junagadh district, were selected for the study. The villages are situated in the heart of Gir forests and are known to be the abode of the Siddis, an Afro-Indian tribal group of Gujarat.

The Siddis are a unique tribe that has African ancestry and survives in South Asia. In India, they are mainly found in three states—Gujarat, Karnataka, and Andhra Pradesh. Some genetic studies have suggested that they are most closely related to Africans (Shah Anish et al. 2011). The Siddis in Gujarat are nearly all Sunni Muslim in faith; they speak an admixture of Gujarati and Hindi in the Saurashtra region of Gujarat. The Siddis believe that they were part of the group of African slaves who were sold to Indian princely states, and they adopted this culture from Muslim army men who dominated the armies of the sultans. Sunni Muslims follow a patrilineal descent through surnames. The Siddis are known to be an endogamous group, and they believe that the descendants of common founder ancestors can intermarry.

Sample size and data collection

Two hundred and twenty-two females (from the sample of 451 women) provided complete information about their pedigrees up to the fourth generation and therefore were included in the study. Inclusion criteria were based on ethnicity, marital status, and willingness to provide genealogical details and records; whereas incomplete genealogies were excluded. All information was gathered based on structured face-to-face interviews by the researcher, block health officers, and ASHA workers using the local language. To construct pedigrees, information was collected through four generations upwards if the women in question are considered a proband. All data gathered was validated simultaneously with various anthropological methods through genealogical checks, personal observations, and cross-checking from different informants. Medical data was obtained from families and was confirmed through medical charts or with the help of medical staff wherever possible. Socioeconomic status was assessed using standard of living index (SLI) applicable for rural India (Davey Smith et al. 2003). Informed written consent or thumb impressions (for non-literates) were obtained from each subject after explaining the objectives of the study in local language. Ethical clearance was obtained as per rules.

Quantification of socioeconomic status

The information on the background characteristics of households such as religion and caste as well as residence was collected. Each household was then assigned an SLI code based on housing characteristics and ownership of assets. On the basis of these, each household was classified into low, medium, and high standard of living (SLI) categories. All individuals in the same household are assigned the same SLI category. The SLI is an alternative standard of living index, calculated using a different method of weighting the indices. A proportionate possession weighting (PPW) is an adjustment that reflects the differences between various social

and demographic groups and therefore takes account of these differences within the population (rural or urban). Unlike the National Family Health Survey (NFHS) standard of living index, this PPW-SLI index refers entirely to a household's possessions. For example, proportionate index weight would allow for differences in the various levels of possession of telephones and tables between urban and rural households. The SLI as a socioeconomic index has been found to be reliable, valid, and usable in India (Prakasam; Subramanian et al. 2006). The PPW based on the NFHS All India 1998 Report used in the study are given in Table 1.

Analysis and statistical measures

The relationship between the spouses was recorded as whether their parents were consanguineous and if either set of grandparents practiced consanguinity. The data was analyzed using the Web inbreeding calculator for calculating p , F , and R (2011; Subramanian et al. 2006; Prakasam). p is the probability that the person is autozygous for an allele with respect to the whole pedigree; F is the person's coefficient of inbreeding with respect to the whole pedigree; while R is the coefficient of relationship for the person, with respect to the whole. Consanguineous marriages were classified in five groups:

double first cousins, first cousins; first cousin once removed; second cousin; less than second cousin (third cousin); and non-consanguineous marriage. Cardio-metabolic health was defined by assessing the occurrence of heart diseases, diabetes, and hypertension. Reported eye defects and problems (myopia, hypermetropia, weak eye muscles, watering eyes, and headaches have also been associated with weak eyes) were high in number and hence categorized separately. Other diseases included blood disorders, kidney problems, hearing defects, asthma, etc.

Chi-square was used to ascertain the association between two or more categorical variables. For a small sample size, Fisher's exact test (two-tailed) was used. Odds ratios were computed for the likelihood of disease by consanguinity status in the current generation as well as the proband's parental generation. Potential effect modification was assessed in the relationship between the risk score and consanguineous status by health status, education, SLI, and age through regression analysis. Cross products were added, one at a time, to fully adjusted models; no cross products were statistically significant. Consanguinity in either set of grandparents (maternal or paternal) was registered for these analyses. Cases were defined as the progeny of consanguineous unions with a disease (either self or siblings), and controls were defined as the progeny of non-consanguineous unions with a disease (either self or siblings). The data was analyzed in IBM SPSS 19.0.

Table 1 Proportionate possession weighting based on the NFHS All India 1998 Report used in the study

Item	All India total		All India urban		All India rural	
	%	Weight	%	Weight	%	Weight
1 Mattress	47.4	52.6	71.7	28.3	38.1	61.9
2 Pressure cooker	29.6	70.4	65.2	34.8	16.0	84.0
3 Chair	45.5	54.5	71.3	28.7	35.6	64.4
4 Cot/bed	81.2	18.8	86.1	13.9	79.4	20.6
5 Table	39.6	60.4	64.9	35.1	30.0	70.0
6 Clock/watch	66.5	33.5	90.1	9.9	57.5	42.5
7 Electric fan	45.5	54.5	82.2	17.8	31.4	68.6
8 Bicycle	47.8	52.2	53.5	46.5	45.7	54.3
9 Radio	38.0	62.0	53.2	46.8	32.2	67.8
10 Sewing machine	18.4	81.6	35.5	64.5	11.9	88.1
11 Telephone	7.4	92.6	20.1	79.9	2.6	97.4
12 Refrigerator	10.6	89.4	28.8	71.2	3.7	96.3
13 TV (B/W)	24.7	75.3	44.8	55.2	17.0	83.0
14 TV (color)	10.1	89.9	27.3	72.7	3.5	96.5
15 Moped	11.2	88.8	25.0	75.0	6.0	94.0
16 Car	1.6	98.4	4.4	95.6	0.6	99.4
17 Tractor	1.6	98.4	0.8	99.2	2.0	98.0
18 Bullock cart	7.2	92.8	1.4	98.6	9.4	90.6
19 Water pump	8.5	91.5	9.3	90.7	8.2	91.8
20 Thresher	2.0	98.0	0.7	99.3	2.5	97.5

Results

Sociodemographic characteristics for consanguineous and non-consanguineous distribution in the study population are shown in Table 2. A significant association was observed between women's age, educational level, occupational level, and consanguinity. Older women (>45 years) were categorized under consanguineous marriages (78.9 %) as opposed to the youngest group where such unions were minimal (30.8 %). More illiterate women were reported of being in consanguineous unions (76.3 %) unlike women with university education who were less likely to be married to cousins (11.1 %). Though the majority of women in consanguineous marriage were manual laborers and those in non-consanguineous marriage were housewives, yet no significant association pattern was seen with the type of unions. Socioeconomic status and consanguinity showed U-shaped associations as the highest consanguineous unions were reported among women with SLI in the two groups: SLI<300 and those with SLI>900. Fifty seven to fifty eight percent of consanguineous women were seen to be significantly associated with consanguinity in parents and in affinal parents.

Table 3 shows trends of consanguinity in the proband's (current) generation compared to the parental generation and

Table 2 Sociodemographic characteristics of consanguineous and non-consanguineous distribution in the study population (expressed in percentages)

Variables		Type of marriage		<i>p</i> value
		Consanguineous (<i>N</i> =109)	Non-consanguineous (<i>N</i> =113)	
Age groups (years)	<25	30.80	69.20	0.001
	25–35	50.60	49.40	
	35–45	62.80	37.20	
	>45	88.90	11.10	
Self-education	Illiterate	76.30	23.70	0.001
	Schooling	63.50	36.50	
	10+2	34.80	65.20	
	Graduate	11.10	88.90	
Occupation	Housewife	43.50	56.50	NS
	Manual	53.20	46.80	
	Others	52.40	47.60	
Standard of living index	<300	63.00	37.00	0.001
	300–600	32.70	67.30	
	600–900	49.80	50.20	
	>900	87.00	13.00	
Consanguinity in parents (of the proband)	Yes	59.34	40.95	0.005
	No	40.17	59.82	
Consanguinity in husband's parents	Yes	57.44	42.55	0.011
	No	40.62	59.38	

grandparent generation with *p*, *F*, and *R* coefficients. The rate of consanguinity in the present generation was 49 % (*N*=109), higher than preceding generations. The rate of consanguinity in the parental generation was similar to that in the current generation (47.3 %), whereas consanguinity rate among the proband's husband's parents was 42.34 %. The most common type of consanguineous marriage is the first cousin marriage in the all generations, followed by unions with first cousin once removed. The coefficients of inbreeding in the proband's, proband's parents, and husband's parents were 0.0316, 0.0219, and 0.0182, respectively, and the average

rates of probability of autozygosity for an allele were 3, 2.2, and 1.6 %, respectively. The coefficient of relationship was highest in the current generation.

The prevalence of cardio-metabolic diseases and other common adult problems among parents (based on marriage pattern of the grandparents) and the current generation by consanguineous versus non-consanguineous mating in the Siddi population is presented in Table 4. Most reported diseases were more frequent in consanguineous unions, and the differences between the two groups for most disease variable results were statistically significant. Nearly three times odds

Table 3 Consanguinity in the proband's (current) generation compared to the parental generation and grandparent generation with *p*, *F*, and *R* coefficients

Degree of consanguinity	Current generation	Parents of the proband	Parents of the proband's husband	Proband's maternal grandparents	Proband's paternal grandparents
Consanguinity	109 (49.10)	105 (47.30)	94 (42.34)	76 (34.23)	70 (35.59)
No consanguinity	113 (50.90)	117 (52.70)	128 (57.66)	138 (65.77)	143 (64.41)
Double cross-cousin	5.04 % (6)	3.81 % (4)	4.26 % (4)		
First cousin	44.04 % (48)	38.10 % (40)	39.36 % (37)		
First cousin once removed	24.77 % (29)	20.00 % (21)	15.96 % (15)		
Second cousin	8.25 % (9)	21.90 % (23)	21.28 % (20)		
Less than second cousin	15.60 % (17)	16.19 % (17)	19.15 % (18)		
<i>p</i>	0.0298	0.0229	0.0166		
<i>F</i>	0.0316	0.0219	0.0182		
<i>R</i>	0.0911	0.0712	0.0664		

Table 4 Prevalence and ORs for cardio-metabolic diseases and other common adult problems among two generations in the Siddi population

Variables		Consanguineous marriage	Non-consanguineous marriage	Chi-square	Exp (<i>B</i>)	CIs		<i>p</i> value
						Lower	Upper	
Parent generation (generation 2)								
Heart diseases	Yes	20	8	15.8***	2.65	1.02	6.85	0.044
	No	66	128					
Diabetes	Yes	21	16	6.07*	2.44	1.26	4.76	0.009
	No	65	120					
Hypertension	Yes	32	22	12.66***	2.62	1.39	4.94	0.003
	No	54	114					
Eye defects	Yes	33	13	26.66***	2.75	1.41	5.35	0.003
	No	53	123					
Other problems	Yes	21	25	ns	1.61	0.78	3.34	ns
	No	65	111					
Present generation (generation 1)								
Heart diseases	Yes	10	6	ns	0.62	0.24	1.65	ns
	No	95	111					
Diabetes	Yes	9	4	6.57**	3.67	1.81	7.44	0.001
	No	96	113					
Hypertension	Yes	18	8	5.68*	2.15	1.12	3.92	0.02
	No	87	109					
Eye defects	Yes	20	8	4.29*	1.54	1.03	1.97	0.01
	No	85	109					
Other problems ^a	Yes	15	9	ns	1.80	0.88	3.69	ns
	No	90	108					

All beta values have been controlled for other disease variables through regression analysis with health status, education, SLI, and age

* $p < 0.05$; ** $p < 0.01$; *** $p < 0.001$

^a Other diseases included blood disorders, kidney problems, hearing defects, asthma, etc.

for cardio-metabolic risks (2.65 odds ratio (OR) for heart diseases, 2.44 OR for diabetes mellitus, and 2.62 OR for hypertension) have been contracted in the progeny of consanguineous marriage in generation 2. Similarly, the current generation (generation 1) of consanguineous parents had a significantly higher risk than the non-consanguineous parents for diseases such as diabetes mellitus (3.67 OR, confidence interval (CI) 1.81–7.44), hypertension (2.15 OR, CI 1.12–3.92), and eye defects (1.54 OR, CI 1.03–1.97).

Discussion

A fundamental unanswered query in anthropological research is the relative importance of biological versus social factors in determining health status. A common biogenetic factor, consanguinity, which is identified as a basic factor of micro-evolution in populations, causes the expression of deleterious recessive genes. Yet, such marriages are favored by different populations usually bound to traditional customs and beliefs and to keep property in a united form within the family (Bittles et al. 1991).

The incidence of consanguinity is relatively high in the present generation as compared to previous generations (49.10 % vs. 47.30 and 42.34 %). The reason for high rates of consanguinity might be cultural beliefs, social life, customs associated with religion (Islam for the present study), and ethnicity which is a way of life in most populations. Also, education, socioeconomic status, consanguinity in parents, and age are important determinants of consanguinity. Among the major populations so far studied, the highest rates of consanguineous marriage have been associated with low socioeconomic status, illiteracy, and rural residence. It was also argued that socioeconomic and cultural factors need to be controlled in order to assess the effects of inbreeding (Bittles 1994).

Another reason for preference of consanguineous marriages in the present generation is the growing concern about the future in terms of long-term marital concordance and financial stability or insurances. It is known that consanguineous marriages are primarily social. In communities with high consanguinity rates, it has been indicated that consanguineous marriage could enforce the couple's stability due to higher compatibility between husband and wife who share the same social relationships after marriage as before marriage, as well

as the compatibility between the couple and other family members. Also, these marriages seem to elevate women's status and reduce the possibilities of hidden uncertainties in health and financial issues (Hamamy 2012). These marriages come with an underlying insurance of non-division of financial resources associated with a family, in later years. Higher consanguineous marriage in the youngest group seems to be a coping mechanism with concerning relatively new social and monetary issues.

The Encyclopedia of Genetics, Genomics, Proteomics, and Informatics suggests that inbreeding coefficient measures the probability that two alleles at a locus in an individual are identical by descent from a common ancestor, i.e., the chance that an individual is homozygous for an ancestral allele by inheritance (not by mutation) (Rédei 2008). The coefficient of inbreeding is highest in the present generation (0.0316), and then proband's parents (0.0219), and finally husband's parents (0.0182). This is due to the fact that cross-cousin marriages were highest in the present generation. Many studies have proved that the coefficients of inbreeding with regard to first cousin and uncle-niece progenies are much higher when compared to that of second cousins, and second cousin progenies are closer to that of non-consanguineous groups (Padmadas and Nair 2001). Imaizumi (1988) also revealed similar results documenting that kinship decreases with marital distance in the parental generation, and socioeconomic class (level of education and occupation) effects are small by comparison with those of the present generation. The rate of consanguinity is significantly higher in the older generation (Imaizumi 1988).

The main impact of inbreeding is an increase in the rate of homozygotes for recessive disorders (Bener et al. 2007), and it has also been reported that several genetic disorders and reproductive wastage are more frequent in consanguineous marriages (Bittles et al. 1991). The impact of consanguinity has been clearly documented in the present studies. The risk of cardio-metabolic disorders was estimated to two to three times in the progeny of consanguineous unions as against non-consanguineous marriages. A greater inbreeding coefficient is directly proportional to a higher risk outcome, though the studies suggest otherwise for heart diseases and other disease variables. This is due to the fact that not all diseases have still been expressed in the current generation which may arise later in life.

Limitations

It is noteworthy that pedigree-based estimates of consanguinity have several limitations; in particular, they do not provide information about close kin marriages that occurred in distant generations. Thus, we might have underestimated the cumulative inbreeding effects, and also, mostly incorrectly ascribed paternity is not recorded. Furthermore, the bias in data

associated with the ages, reporting of diseases, and marriage pattern due to underreporting or misreporting cannot be ruled out.

Conclusion

The risk of cardio-metabolic diseases is higher in offspring of consanguineous couples. If contextually expressed, it can also be concluded that in a population with a high rate of consanguinity, there is a significant increase in the prevalence of common adult diseases.

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Compliance with ethics guidelines The experiments performed in this study comply with the current laws of the country in which they were performed.

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