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Consanguinity as a Determinant of Reproductive Behaviour and Mortality in Pakistan

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To determine the prevalence of consanguineous marriages and estimate the effects of consanguinity on reproductive behaviour and mortality, household and hospital-based surveys were conducted in 11 cities in the Pakistan province of Punjab between 1979 and 1985. The 9620 women interviewed reported 44 474 pregnancies, with data collected on maternal and paternal ages at marriage, abortions/miscarriages, stillbirths and deaths in the first month, at 2–12 months and 2–8/10 years. Six categories of consanguineous marriage were included: double first cousin, first cousin, first cousin once removed/double second cousin, second cousin, *bradari* (brotherhood) and non-consanguineous. Marriages contracted between spouses related as second cousins or closer accounted for 50.3% of the total, equivalent to an average coefficient of kinship ($\alpha = \Sigma p_i F_i$) of 0.0280. Unions between close biological relatives were characterized by younger maternal and paternal ages at marriage and reduced spousal age difference, but a longer time to first delivery. Overall, they exhibited greater fertility than non-consanguineous couples. Antenatal and postnatal mortality were assessed by consanguinity and age interval. Consanguinity-associated deaths were consistently higher in the neonatal, infant and childhood periods. The consequences of these outcomes on the health of the present and future generations is assessed.

Consanguinity is a central feature of kinship systems in many parts of the world, including North and sub-Saharan Africa and Western, Central and Southern Asia, where marriages between close biological relatives account for 20–55% of all unions.[1] In general, the highest rates of consanguineous marriage are reported in more traditional rural areas and among the poorest and least educated sections of society.[2,3] However in Japan[4] and South India,[5] preferential marriage between close relatives is also favoured by major land-owning groups. In both cases the maintenance of family property is a major consideration with only token or significantly reduced dowry and bridewealth payments required,[6,7] but among land-owners the preservation of estates and land-holdings is probably the critical factor. Besides these economic considerations, consanguineous marriages are preferred because of the comparative ease with which prenuptial negotiations can be conducted. It also is believed that such unions offer the optimum opportunity for compatibility, both between husband and wife and the bride and her mother-in-law,[8] thus benefiting female status.[9,10] Perhaps of greatest importance, there is the underlying conviction that, by marrying within the extended family, hidden uncertainties regarding health or other unfavourable family characteristics will not arise.[11]

With the exception of Hindus and Sikhs in North India, who practise caste endogamy but strenuously prohibit marriage to a biological relative,[12] consanguineous marriages have been widely reported in all other major religious groups of Southern Asia, including South Indian Hindus.[2] The preferred form of consanguineous union varies by religion, and to a lesser extent by region. Thus, although uncle-niece unions ($F = 0.125$) are common among South Indian Hindus, they are proscribed by the Koran which nevertheless permits double first cousin unions ($F = 0.125$). First cousin marriages predominate among Muslims, particularly father's brother's daughter, with the couple on average sharing identical gene copies inherited from a common ancestor(s) at one-eighth of all loci. As a result their progeny would be homozygous at one-sixteenth of all autosomal loci, expressed mathematically as the coefficient of inbreeding (F) = 0.0625. The aim of the present study was to assess the effects of inbreeding within urban populations in Pakistan,

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with particular emphasis on reproductive behaviour and postnatal mortality.

SUBJECTS AND METHODS

Data on 9520 families resident in 11 cities in the province of Punjab, Pakistan were obtained in studies conducted between 1979 and 1985. All information was based on retrospective interview in the respondents' own language. For Lahore, and in part Gujrat, the women were interviewed as obstetric inpatients, with data from the remaining nine localities collected by household interview. The cities surveyed ranged in population size from 49 500 (Mianchannu) to 3.8 million (Lahore). Data categories included the age of both partners at marriage, spousal age difference, time to first delivery, number of pregnancies, number of abortions/miscarriages, stillbirths, deaths in the first month of life, at 2–12 months and 2–8/10 years.

Information was also obtained on the biological relationship between husband and wife. As a working definition, unions contracted between people related as second cousins or closer were categorized as consanguineous. These arbitrary limits were chosen because the genetic influence in marriages between couples related to a lesser degree would be expected to differ only marginally from that observed in non-consanguineous unions. The consanguinity classes recorded were double first cousin (equivalent to a coefficient of inbreeding in the progeny of $F = 0.125$), first cousin ($F = 0.0625$), first cousin once removed/double second cousin ($F = 0.0313$), second cousin ($F = 0.0156$), *bradari* ($F < 0.0156$) and non-consanguineous ($F = 0$). The term *bradari* literally translates as brotherhood, and in Pakistan it is used to denote relationships along fraternal and/or paternal lines. Because of the imprecise definition of these marital unions with respect to consanguinity, *bradari* marriages were treated as non-consanguineous when calculating percentage inbreeding. However, as in all endogamous societies with breeding pools of finite size, some degree of random inbreeding would be expected in the relationships. The average levels of inbreeding were assessed in terms of coefficient of kinship values for each population ($\alpha = \Sigma p_i F_i$), where α measures the probability that a gene taken at random from one spouse is identical by descent to a gene from the same locus taken at random from their partner.

To investigate the relationships between consanguinity, fertility and pre-reproductive mortality, mean maternal and paternal age at marriage, spousal age difference, time to first delivery, number of pregnancies and livebirths, and mortality by age interval, were calculated for each consanguinity class using the Genstat 5 statistical package (Rothamstead Experimental Station).

RESULTS

Of the 9520 marriages investigated, 50.25% were contracted between spouses related as second cousins or closer, equivalent to an average coefficient of kinship for the population ($\alpha = \Sigma p_i F_i$) of 0.0280 (Table 1). First cousin marriages ($F = 0.0625$), in which the partners have one set of grandparents in common, were the most common form of consanguineous union (37.07%), with 0.90% double first cousins, 11.72% first cousin once removed/double second cousins and 0.56% second cousin unions. In Punjab, the high reported ratio of first cousin to second cousin marriage can most convincingly be ascribed to the strength of preference for the former marital pattern in this strongly Islamic society. A further 33.93% of marriages were *bradari* relationships. The percentage consanguinity and coefficients of kinship both indicated that the prevalence and types of inbred union were similar across localities. Since the consanguinity classifications refer to the present generation only, i.e. more distant common ancestors than great-grandparents were not considered, the average α values calculated for each locality represent minimal inbreeding estimates for these communities.

Maternal and paternal ages at marriage were negatively associated with consanguinity, for example, averaging 18.35 and 23.28 years in double first cousin unions, by comparison with 19.74 and 25.97 years in non-consanguineous couples (Table 2). Spousal age differences were also negatively associated with consanguinity, increasing from 4.51 years in double first cousins to 6.54 years in the non-consanguineous group, but there was a positive association between consanguinity and time to first delivery. There was a positive association between consanguinity and mean number of pregnancies (Table 3). This relationship persisted for livebirths, although the differential was reduced because of greater antenatal losses in consanguineous marriages. Total pre-reproductive mortality also increased with degree of inbreeding (Table 4). As a percentage of all reported pregnancies, total ante- and postnatal mortality ($\leqslant 10$ years) rose from 16.36% in non-consanguineous progeny, to 20.09% in second cousins, 22.91% in first cousins once removed/double first cousins, 22.13% in first cousins and 38.97% in double first cousins. The mortality differentials were lowest in reported abortions/miscarriages, and exerted proportionately greatest effect during the first postnatal month.

TABLE 1 *Data source, number of marriages studied, percentage consanguinity, and average coefficient of kinship (α) by locality*

Locality	Data source	No.	Percentage[a] consanguinity	Coefficient of kinship (α)
Lahore	Obstetric inpatients	966	47.20	0.0269
Sheikhupura	Household survey	1007	48.86	0.0271
Muridke	Household survey	251	40.24	0.0240
Mianchannu	Household survey	135	37.78	0.0236
Gujrat	Obstetric inpatients/ household survey	1002	48.50	0.0277
Rawalpindi	Household survey	1000	48.10	0.0286
Jhelum	Household survey	1027	44.30	0.0262
Gujranwala	Household survey	1059	58.92	0.0323
Sahiwal	Household survey	1003	56.13	0.0295
Faisalabad	Household survey	1033	52.08	0.0293
Sialkot	Household survey	1037	51.78	0.0261
Punjab[b]	—	9520	50.25	0.0280

[a] Includes all marriages at the level of second cousin ($F = 0.0156$) or closer.
[b] Aggregate data for all 11 cities.

TABLE 2 *Mean (and standard error) of parental age at marriage in years, spousal age difference, and time to first delivery, by consanguinity class*

	D1C[a]	1C[b]	1.5C/D2C[c]	2C[d]	*Bradari*[e]	NC[f]
Maternal age	18.35 (1.37)	18.79 (0.70)	18.71 (0.71)	20.03 (1.47)	19.01 (0.77)	19.74 (0.88)
Paternal age	23.28 (0.96)	23.13 (0.92)	23.53 (0.84)	25.31 (2.31)	24.06 (1.01)	25.97 (1.82)
Spousal age difference	4.51 (1.97)	4.62 (0.86)	4.92 (1.25)	5.46 (2.78)	5.26 (1.17)	6.54 (1.30)
Time to first delivery	2.06 (0.89)	1.91 (0.35)	1.91 (0.27)	1.28 (0.36)	1.76 (0.35)	1.58 (0.38)
No. of marriages	86	3529	1116	53	3230	1506

[a] D1C, double first cousin ($F = 0.125$).
[b] 1C, first cousin ($F = 0.0625$).
[c] 1.5C/D2C, first cousin once removed/double second cousin ($F = 0.0313$).
[d] 2C, second cousin ($F = 0.0156$).
[e] *Bradari*, ($F < 0.0156$).
[f] NC, non-consanguineous ($F = 0$).

TABLE 3 *Total pregnancies and livebirths per mother by consanguinity class*[a]

	D1C	1C	1.5C/D2C	2C	*Bradari*	NC
Pregnancies	4.95	4.75	4.70	4.13	4.77	4.32
Livebirths	4.23	4.37	4.35	3.83	4.47	4.02
No. of marriages	86	3529	1116	53	3230	1506

[a] See footnote Table 2.

TABLE 4 *Proportion of deaths by age interval and consanguinity class*[a]

	DIC	1C	1.5C/D2C	2C	*Bradari*	NC
Abortions/miscarriages	0.0563	0.0405	0.0410	0.0365	0.0320	0.0438
Stillbirths	0.0892	0.0380	0.0336	0.0365	0.0258	0.0269
First month	0.0516	0.0345	0.0372	0.0320	0.0252	0.0207
2–12 months	0.0751	0.0447	0.0551	0.0365	0.0399	0.0301
2–8/10 years	0.1174	0.0635	0.0622	0.0594	0.0559	0.0421
Total mortality	0.3897	0.2213	0.2291	0.2009	0.1788	0.1636
No. of pregnancies	426	16754	5242	219	15325	6508

[a] See footnote Table 2.

DISCUSSION

Despite earlier predictions of a marked decline in the frequency of consanguineous unions in Southern Asia,[13] studies in South India during the 1980s have confirmed the continuing popularity of this form of marriage.[14] Preliminary analysis of the 1990–1991 Pakistan Demographic and Health Survey (PDHS) has shown that marriage to a close biological relative is favoured in all provinces, but with significantly larger numbers of inbred unions in rural areas.[15] For urban Punjab populations α was 0.0292, very close to the reported figure in the present study, whereas the equivalent value calculated from the PDHS data for rural areas was $\alpha = 0.0371$. In Punjab a study on *qaums*, endogamous groupings traditionally identified by occupation, indicated that although there were variations in the prevalence of consanguinity, the preference for unions between close kin effectively crossed all social/occupational boundaries.[16]

Younger parental age at marriage, in particular younger maternal age (Table 2), is a significant feature of consanguineous unions, since it facilitates increased levels of fertility by optimizing maternal reproductive span and concentrating childbearing in the most fertile years. The importance of this factor in determining family size is seen in the higher net fertility of consanguineous couples (Table 3), in spite of their generally longer time to first delivery and greater levels of ante- and postnatal mortality. Unfortunately information was not collected on birth intervals, and so it is not possible to determine whether the greater fertility in consanguineous unions also could be linked to reproductive compensation, as suggested in Japan.[4]

The positive association between total mortality and degree of consanguinity, and the timing of deaths with greatest effect in the first year (Table 4), strongly suggest that the expression of deleterious recessive genes was a significant factor in the excess mortality. An associated pilot study has further indicated that this disadvantage may extend into adult morbidity.[17] In the absence of adequate socioeconomic control the results must be treated with caution, particularly with respect to the very high rates of neonatal deaths among double first cousin progeny, when young maternal age may have been an important contributory factor.[18,19] Conversely, it should be remembered that in many populations where consanguinity is the preferred form of marital union, data on postnatal mortality commonly have been assessed solely in terms of socio-demographic variables, such as maternal education, birth order and birth interval and without reference to possible detrimental effects of inbreeding.

Preliminary information from the PDHS[15] indicate the existence of socioeconomic differentials between consanguineous and non-consanguineous families, with unrelated couples occupying the more favoured socioeconomic position. However, there appear to be few differences between the two groups in terms of their access to and uptake of vaccination for maternal tetanus and common childhood infectious diseases. National studies on the large Pakistani community in the UK, with a first cousin marriage rate of approximately 55%,[20] confirm their high levels of perinatal and infant mortality across all socioeconomic classes, and congenital anomalies have been identified as a major cause of infant death.[21] Similarly, an ongoing prospective investigation in the British Midlands has further demonstrated the significant relationship between consanguinity and the incidence of major congenital malformations and specific autosomal recessive disorders in locally-resident Pakistanis.[22,23]

Many of the interpretative problems in differentiating between genetic and non-genetic causes of premature mortality would be greatly assisted by comprehensive biosocial studies. Until such data are available it is appropriate to emphasize that in

Pakistan, and among overseas Pakistani communities, consanguineous unions are judged to offer significant social, economic and cultural benefits. Improved public health measures, including greater provision of vaccination for childhood infectious diseases, should enable larger numbers of children to survive to marriageable age. In turn, this would permit an even greater proportion of the population to exercise their preference for marriage to a close biological relative. But with development of the country, and a decline in mortality due to infectious and nutritional diseases, an equivalent increase in the proportion of people diagnosed with genetic disorders can be predicted, a trend already apparent in the UK Pakistani community. Based on UK experience to date, planning for this major transition in the disease profiles of many of the more populous developing countries should be instituted without delay.

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REFERENCES

[1] Bittles A H. *Consanguineous Marriage: Current Global Incidence and its Relevance to Demographic Research*. Population Studies Center, Research Report No. 90–186. Ann Arbor: University of Michigan, 1990.

[2] Bittles A H, Mason W M, Greene J, Appaji Rao N. Reproductive behavior and health in consanguineous marriages. *Science* 1991; **252**: 789–94.

[3] Bittles A H. Consanguinity: a major variable in studies on North African reproductive behaviour, morbidity and mortality? In *Proceedings of the Demographic and Health Surveys World Conference*. Moore S (ed.). Institute for Resource Development, Columbia, MD, 1992, vol. 1, 321–41.

[4] Schull W J, Neel J V. The effects of parental consanguinity and inbreeding in Hirado, Japan. V. Summary and interpretation. *Am J Hum Genet* 1972; **24**: 425–53.

[5] Rami Reddy V, Chandrasekhar Reddy B K. Consanguinity effects on fertility and mortality among the Reddis of Chittoor District (A.P.) South India. *Ind J Hered* 1979; **11**: 77–88.

[6] Caldwell J C, Reddy P H, Caldwell P. *The Causes of Demographic Change: Experimental Research in South India*. Madison: University of Wisconsin Press, 1988, p. 239.

[7] Govinda Reddy P. Consanguineous marriages and marriage payment: a study among three South Indian caste groups. *Ann Hum Biol* 1988; **15**: 263–68.

[8] Dronamraju K R, Meera Khan P. The frequency and effects of consanguineous marriages in Andhra Pradesh. *J Genet* 1963; **58**: 387–401.

[9] Dyson T, Moore M. On kinship structure, female autonomy and demographic behavior in India. *Pop Dev Rev* 1983; **9**: 35–59.

[10] Bittles A H, Mason W M, Singarayer D N, Shreeniwas S, Spinar M. Determinants of the sex ratio in India: studies at national state, and local levels. In: Schell L, Smith M T, Bilsborough A (eds). *Urban Ecology and Health in the Third World*. Cambridge: Cambridge University Press, 1993, pp. 244–59.

[11] Bittles A H, Shami S A, Appaji Rao N. Consanguineous marriage in South Asia: incidence, causes, and effects. In: Bittles A H, Roberts D F (eds). *Minority Populations: Genetics, Demography and Health*. London: Macmillan, 1992, pp. 102–17.

[12] Kapadia K M. *Marriage and Family in India*. Calcutta: Oxford University Press, 1958, p. 117.

[13] Haldane J B S. The implications of genetics for human society. In *Genetics Today: Proceedings of the 11th International Congress of Genetics*. Geerts S J (ed). Pergamon Press, The Hague, 1963, pp. 91–102.

[14] Bittles A H, Coble J M, Appaji Rao N. Trends in consanguineous marriage in Karnataka, South India, 1980–1989. *J Biosoc Sci* 1993; **25**: 111–16.

[15] Ahmed T, Ali S M, Aliaga A *et al*. *Pakistan Demographic and Health Survey 1990/91*. Islamabad: National Institute for Population Studies, and Columbia, Maryland: Institute for Resource Development, 1992.

[16] Shami S A, Grant J C, Bittles A H. Consanguineous marriage and social/occupational class boundaries in Pakistan. *J Biosoc Sci* 1993; (In press).

[17] Shami S A, Qaisar R, Bittles A H. Consanguinity and adult morbidity in Pakistan. *Lancet* 1991; **338**: 954–55.

[18] Shami S A, Schmitt L H, Bittles A H. Consanguinity-related prenatal and postnatal mortality in the populations of seven Pakistani cities. *J Med Genet* 1989; **26**: 267–71.

[19] Shami S A, Schmitt L H, Bittles A H. Consanguinity, spousal age at marriage and fertility in seven Pakistani Punjab cities. *Ann Hum Biol* 1990; **17**: 97–105.

[20] Modell B. Social and genetic implications of customary consanguineous marriage among British Pakistanis. *J Med Genet* 1991; **28**: 720–23.

[21] Balarajan R, Soni Raleigh V. A health profile of Britain's ethnic minorities. In: Bittles A H, Roberts D F (eds). *Minority populations: Genetics, Demography and Health*. London: Macmillan, 1992, pp. 119–42.

[22] Bundey S, Alam H, Kaur A, Mir S, Lancashire R. Why do UK-born Pakistani babies have high perinatal and neonatal rates? *Paed Perinat Epidemiol* 1991; **5**: 101–14.

[23] Bundey S. A prospective study on the health of Birmingham babies in different ethnic groups: interim findings. In: Bittles A H, Roberts D F (eds). *Minority Populations: Genetics, Demography and Health*. London: Macmillan, 1992, pp. 143–55.

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