CONSANGUINITY AND ITS EFFECT ON OFFSPRING IN AN URBAN COMMUNITY OF GUNTUR CITY OF ANDHRA PRADESH

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ABSTRACT

BACKGROUND

While consanguineous marriages enforce family solidarity and reduce the possibilities of hidden uncertainties, studies show that they are critical predictors of adverse pregnancy outcomes. Obstetricians are faced with consanguineous couples anxious to know the anticipated health risks to their offspring. This study is set to describe the magnitude of consanguinity in an urban population and to identify the effects of consanguinity in terms of congenital malformations and other health issues.

MATERIALS AND METHODS

This study was conducted in the Nallacheruvu area of Guntur city of Andhra Pradesh from June to September 2016. A house to house survey was done in an urban community of Guntur district and all the women with under-five children were identified. A pre tested self-designed structured questionnaire was administered after obtaining informed consent. Couples biologically related as second cousins or closer were classified as consanguineous. The data collected was entered and analysed in MS excel and is presented in tables. Important findings were subjected to tests of significance like Chi Square test at 5% Level of significance.

RESULTS

Of the 583 women enumerated, 81.3% were in a non-consanguineous marriage and the rest 18.7% were in a consanguineous marriage. Muslim families were less in favour of consanguineous marriages compared to the Hindu and Christian families and this difference is statistically significant (p value 0.009). There were no instances of consanguineous marriages among the ST families. It was seen that larger families; had a higher level of consanguinity. More of the children born to consanguineous parents were having chronic illnesses (7.5%) compared to the nonconsanguineous (4.1%). Number of episodes of illness in the year was found be more in the children of consanguineous parentage (p 0.03).

CONCLUSION

The effect of consanguinity in terms of child morbidity is a real threat in our communities. The establishment of more population and hospital based registries will help identify the true prevalence of congenital malformations due to consanguinity. Premarital genetic counseling or counseling prior to conception should be made freely available.

KEYWORDS

Consanguineous, Congenital Defects, Chronic Illnesses, Illness Episodes.

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BACKGROUND

In many communities, consanguineous marriages offer social and economic advantages and also strengthening of family relationships. These compensations often outweigh the biological disadvantages of close-kin marriage for a majority of families.[1] From the woman's point of view, consanguineous marriage allows a better relationship with her in-laws who could support her in time of need. Generally speaking, marrying within the family reduces the possibilities of hidden uncertainties in health and financial issues. Consanguinity strengthens family ties and enforces family solidarity. These marriages also provide excellent opportunities for the transmission of cultural values and cultural continuity.[2]

Globally it is estimated that at least 8.5% of children have consanguineous parents.[3] Kuntla et al state that the overall prevalence of consanguinity in India is around 16% (1- 4% in the northern region to as high as 40-50% in the southern region).[4] Marriages contracted between persons biologically related as second cousins or closer are categorized as consanguineous. In marriages between married couples related to a lesser degree, the genetic influence on the progeny would usually differ only slightly from that in the general population.

Structural or functional abnormalities such as birth defects or disorders that occur during intrauterine life are considered as congenital anomalies. They may be identified

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prenatally, at birth, or sometimes in case of hearing defects only later in infancy. Major congenital malformations are conditions of prenatal origin which are potentially life threatening. If not corrected, they would impair the child's development or well-being. Minor ones are those which affect non-vital organs and have little or no functional effect. They do not cause distress in the neonatal period.[5] Kuntla S et al found that the relative risk of having stillbirth among consanguineous groups is 1.59, miscarriage is 1.94 and abortion is 3.03 indicating that consanguineous marriages are critical predictors of adverse pregnancy outcomes in Indians.[4]

Bittles AH et al found that in first cousin marriages fertility rate is slightly higher, abortion rate is not different, stillbirths and infant mortality rates are slightly higher. Birth defects frequency was estimated to be around 4-6%. They suggest that consanguineous unions lead to increased expression of autosomal recessive disorders.[6] Hamamy H et al state that the closer the biological relationship between parents, the greater is the probability that their offspring will inherit identical copies of one or more detrimental recessive genes. If parents are unrelated, their chance of having a child with a birth defect or disability is between 2% and 3%. If parents are first cousins, the chance is a little higher at 5% to 6%.[7] In a study done in Norway by Magnus P et al it was seen that the mean birth weight was significantly lower (3377 g vs. 3491 g), and the variance in birth weight was slightly larger for children of consanguineous parentage. The stillbirth rate and neonatal death rate was also higher. Malformations detected shortly after birth was 4.6% for consanguineous when compared to the offspring of non-consanguineous parents (2.2%).[8]

Stoll C et al in a study of 131,760 consecutive births in France, suggest that parental consanguinity is a significant cause of increased incidence of birth defects. Consanguineous mothers had more number of pregnancies and more stillbirths when compared to non-consanguineous mothers.[9]

In view of the higher incidence of congenital anomalies in poorer families around the globe, the WHO states that low-income may be an indirect determinant. About 94% of severe congenital anomalies occur in low- and middle-income countries. Other factors which contribute to this increased risk relate to a lack of sufficient nutritious foods by pregnant women, an increased exposure to factors such as infection and alcohol, or poorer access to healthcare. Bhattacharjee AK et al suggest that as India has around 27 million births every year, there is a need for a systematic surveillance for birth defects such as the Birth Defects Registry of India (BDRI) which was established in 2001. Prevalence of birth defects provided by BDRI in 2010 was 84.2/10000 which is much lower than the estimated prevalence of at least 2%.[10]

A chronic condition is defined as a health problem that lasts over three months and affects the child's normal activities. It also requires much hospitalization and home health care. Pal R et al suggest that chronically ill children, and their families, are at greater risk of developing psychological and emotional difficulties.[11] Primary health care providers are faced with consanguineous couples demanding answers to their questions on the anticipated health risks to their offspring. Preconception and premarital counseling on consanguinity should be part of the training of health care providers particularly in highly consanguineous populations.[12]

Aim- To describe the magnitude of consanguinity in an urban population and to identify the effects of consanguinity in terms of congenital malformations.

MATERIALS AND METHODS

This study was conducted in the Nallacheruvu area of Guntur city of Andhra Pradesh with a population of approximately 16,000 from June to September 2016. A house to house survey was done in this largely low income urban community.

a) Inclusion Criteria: all the women with under-five children were identified and included in the study.
b) Exclusion criteria: Women with older children and those who were not available in the house at the time of visit were excluded from the study.

A pre tested self-designed structured questionnaire on socio-demographic profiles, consanguinity in marriage and also congenital malformations in their children was administered to each one after obtaining informed consent. Couples biologically related as second cousins or closer were classified as consanguineous. The data collected was entered and analysed in MS excel and is presented in tables and graphs. Important findings were subjected to tests of significance like Chi Square test at 5% Level of Significance.

RESULTS

In this study, 583 women were enumerated. 474 (81.3%) of the women were in a non-consanguineous marriage and the rest 109 (18.7%) were in a consanguineous marriage. The number of under-five children were 740. Of these 82.0% were from non-consanguineous parentage and 18.0% from consanguineous parentage.

Among mothers of under-five children included in the study, it was seen that consanguineous marriages were slightly more in the younger age group (<25 years). Religion wise, Muslim families were less in favour of consanguineous marriages compared to the Hindu and Christian families and this difference is statistically significant (p value 0.009). There were no instances of consanguineous marriages among the ST (Scheduled tribe) families. There was no difference in the consanguinity patterns concerning socio-economic status, gender of the baby and educational levels of the women. However, it is seen that larger families; especially the joint families had a higher level of consanguinity. (Table 1)

Sl. No.	Variable	Non-Consanguineous (n=607)	Consanguineous (n=133)	Chi Square	p value
1.	Gender - Male baby	307 (50.6 %)	60 (45.1 %)	1.3	0.2
2.	Age of mother <25 years	317 (52.2 %)	80 (60.2 %)	2.8	0.09
3.	Religion - Muslims	129 (21.3 %)	13 (9.8 %)	9.29	0.009 **
4.	Caste - ST	13 (2.1%)	Nil		
5.	Socio economic status - Low	275 (45.3 %)	55 (41.4 %)	0.76	0.7
6.	Type of Family - Joint	24 (4.0 %)	11 (8.3 %)	7.24	0.03*
7.	Woman's education - Degree, PG, Prof	178 (29.3 %)	42 (31.6 %)	3.11	0.07

Table 1. Consanguinity and Socio-demographic variables

*Significant, ** Highly significant.

Maternal complications and their relationship to consanguinity such as anaemia, eclampsia or PET, Mal-presentations, Caesarean sections, complications at birth for mother and baby etc. were looked at and no significant differences were found (Table 2).

Sl. No.	Variable	Non-Consanguineous (n=607)	Consanguineous (n=133)	Chi Square	p value
1.	Anaemia	66 (10.9 %)	16 (12.0 %)	0.15	0.7
2.	PET / Eclampsia	17 (2.8 %)	7 (5.3 %)	2.11	0.1
3.	Mal presentations	41 (6.8 %)	10 (7.5 %)	0.1	0.8
4.	Type of delivery - LSCS	162 (26.7 %)	40 (30.1 %)	0.63	0.4
5.	Complications at birth - baby	54 (8.9 %)	16 (12.0 %)	1.25	0.3
6.	Complications at birth - mother	219 (36.1 %)	45 (33.8 %)	0.24	0.6

Table 2. Consanguinity and Maternal Complications

Looking at under-five child health related complications like; condition at birth, congenital defects, and delay in physical, mental and social milestones, no significant differences were revealed. More of the children born to consanguineous parents were having chronic illnesses (7.5%) compared to the non-consanguineous (4.1%). Number of episodes of illness in the year was found be more in the children of consanguineous parentage. This finding is statistically significant (p 0.03). (Table 3)

Sl. No.	Variable	Non-Consanguineous (n=607)	Consanguineous (n=133)	Chi Square	p value
1.	Birth condition- Sick	111 (18.3 %)	29 (21.8 %)	0.88	0.4
2.	Congenital defects	11 (1.8 %)	4 (3.0 %)	0.31	0.2
3.	Delayed physical milestones	7 (1.2 %)	Nil		
4.	Delayed mental milestones	7 (1.2 %)	Nil		
5.	Delayed social milestones	4 (0.7 %)	1 (0.8 %)	0.22 (Yates)	0.6
6.	Chronic diseases	25 (4.1 %)	10 (7.5 %)	2.8	0.09
7.	> 4 Episodes of illness in 1 year	7 (1.2 %)	6 (4.5 %)	5.32 (Yates)	**0.02***

Table 3. Consanguinity and Child Complications

*Significant.

Congenital defects, mostly of a minor level, were seen in 3.0% of children of consanguineous parents while it was seen in 1.6% among children of non-consanguineous parentage. However there is no significant difference between the two groups. The overall prevalence of congenital defects in this study is 2.0% with no gender difference. The congenital defects identified are listed in Table 4.

Sl. No.	Defect	Non-Consanguineous (n=607)	Consanguineous (n=133)	Total (n=740)
1.	Polydactyly	1 (0.2 %)	1 (0.8%)	2 (0.3 %)
2.	Club Foot	1 (0.2 %)	1 (0.8%)	2 (0.3 %)
3.	Deaf & Dumb	1 (0.2 %)	1 (0.8%)	2 (0.3 %)
4.	Imperforate anus	1 (0.2 %)	0	1 (0.1 %)
5.	Cleft lip / nasal defect	1 (0.2 %)	1 (0.8%)	2 (0.3 %)
6.	Strabismus	1 (0.2 %)	0	1 (0.1 %)
7.	Hypospadias	1 (0.2 %)	0	1 (0.1 %)
8.	Microcephaly	1 (0.2 %)	0	1 (0.1 %)
9.	Mental retardation	3 (0.5 %)	0	3 (0.4 %)
	Total	11 (1.8 %)	4(3.0%)	15 (2.0 %)

Table 4. List of Congenital Defects

DISCUSSION

Globally it is estimated that at least 8.5% of children have consanguineous parents.[3] In India, the recent estimation of consanguinity rates vary from as low as 1-4% in the northern region to as high as 40-50% in the southern region.[13] Kuntla S et al state that the overall prevalence of consanguinity is found to be 16% which is line with the present findings. Culturally, the Schedules Tribe communities seem to avoid consanguineous marriages while it is less popular among the Muslim communities. Among all the other communities, this practice does not seem to be influenced much by demographic factors such as SES or educational levels.

If the parents are unrelated, their chance of having a child with a birth defect or disability is between 2% and 3%. If parents are first cousins, the chance is a little higher at 5% to 6%. This is due to the increased chance that they will both carry the same autosomal recessive mutation, passed down through the family.[14] Bittles AH et al suggest that in first cousin marriages, fertility rate is slightly higher, abortion rate is not different, stillbirths and infant mortality rates are slightly higher. Birth defects frequency is estimated to be around 2–3% points more than in the general population (around 2–3%). Consanguineous unions lead to increased expression of autosomal recessive disorders.[6]

Mosayebi Z et al found in their study that among the consanguineous group 7.0% of births had congenital anomalies, of which 72.2% were in first-cousin marriages and 27.8% were in second cousin or more distant relatives. Congenital malformations in the non-consanguineous group were 2.0%. They concluded that congenital malformations were 3.5 times more common in consanguineous versus non-consanguineous marriages. This is however not seen in this study where the incidence of congenital defects is much lower.[5]

A study done by PSS Rao et al in South India in 1979 showed that of marriages in rural areas, 46.9% were consanguineous, and in urban areas, 29.1%. In more than 80% of the consanguineous marriages, the spouses were first cousins or more closely related. The findings showed that due to long-term inbreeding there were only marginal or non-significant effects on fertility of inbred populations.[15] Narkhede et al reported 0.25% of mental retardation, 0.5% squint, 0.25% phimosis, 0.25% cleft lip and 0.25% undescended testes.[16]

CONCLUSION

Where there is a high degree of consanguinity, there is a need for public health programs which include prenatal diagnosis, neonatal screening, and genetic counseling. More important is the need for educating the people about the expected genetic fallout. A systematic surveillance system for birth defects in India like the BDRI across the whole country will definitely be of immense help.[17] The establishment of more population and hospital based registries will help identify the true prevalence of congenital malformations due to consanguinity. Premarital genetic counseling or counseling prior to conception should be made available. A non-judgmental attitude towards consanguineous couples is necessary to establish an effective working relationship between the medical profession and communities where consanguineous marriages are prevalent.[18]

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