

Consanguinity and its Effects : The Love That Dare Not Speak Its Surname!

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ABSTRACT

Introduction: Consanguineous marriage means marrying biologically related or blood relatives. Consanguinity is a deeply rooted trend in the South Indian States.

Aims and Objectives : The aim of the study is to determine the current prevalence and pattern of consanguinity, compare and estimate the health parameter in the offspring of consanguineous couples with those of non consanguineous couples, investigate attitudes and awareness of the target population regarding consanguinity and its associated reproductive risk; finally educating the target population, so that they can guide the society well.

Materials & Methods: The study was done in Prathima Institute of Medical Sciences. A population of 500 students between age group of 18-25 years of both sexes, randomly selected from Telangana . A questionnaire was designed which was the key in collecting data from the target population.

Results:

- Prevalance: 41% against 59% non-consanguineous parents..
- Hindus (41.2%) preferred uncle-niece union, Muslim (51.7%) favoured 1st cousins, Christians (54.5%) opted for 2nd cousins.
- Pattern among Hindus: Uncle-niece union 41.5%, first cousins 30.7%, second cousins 15.6%.
- 19% consanguinity presented birth defects; statistically significant.
- 46.1% rural population were consanguineous; statistically significant.
- History of early death / miscarriage / still birth was found in 16.1% of consanguinity; statistically significant .
- Medicos (76.9%) were more aware of consanguinity and its effect compared to paramedical students; is statistically significant.

Conclusion: This study was a preliminary step to obtain first hand information about the magnitude of consanguinity in our state. Appropriate data can guide consanguineous couples take informed decisions regarding reproductive choices.

Keywords: Awareness, Birth defects, Consanguinity, Genetic counseling,

INTRODUCTION

Consanguineous marriage means marrying biologically related or blood relatives. Derived from Latin word "consanguinitas", con meaning shared, sanguine meaning 'of common blood'¹. In clinical genetics, it is defined as a marriage or union between two individuals who are related as second cousins or even closer². It is a deeply rooted social trend in parts of the world like Japan, Middle East & South India². Popularly called as Menarikam in Telangana State . Usually seen in the rural areas, especially in the low socio-economic people, more common among illiterates, or with primary level education². It is estimated that 1 billion(1/5th) of the current global population prefer these marriages^{3,4}. They constitute 20-50% of all marriages, with first cousin unions accounting for almost one-third of all marriages^{3,5,6}. Cognate marriages are culturally and socially favoured as it could enforce the couples stability, as they share the same social relationship before and after the marriage, although there is some recognition of the resulting genetic effects . Widespread consanguinity in a community will lead to relative increase in frequency of affected homozygotes, but a relative decrease in frequency of heterozygotes. Consanguinity increases the chance that both parents carry a gene affected by an identical mutation that they inherited⁷.

This study aims to portray the definition and current trends of consanguinity and prepare simple guidelines for premarital and preconceptive counseling to the health care providers. Because, primary health care providers are confronted with consanguineous couples demanding answers to their questions on the anticipated health risks to their offspring. Preconceptive genetic counseling for consanguinity is considered one of the important pillars amongst the community genetic services in highly consanguineous couples⁵.

Appropriate data can guide health care providers and consanguineous couples take informed decisions regarding their reproductive choices. The present study is a preliminary one to obtain first hand information about the magnitude of the problem in our state. The future prevalence and status of consanguineous marriage is a matter of conjecture.

MATERIALS AND METHODS

Our study is conducted in Prathima Institute of Medical Sciences, Karimnagar, Telangana State.

Specimen selection: Randomly selected subjects hailing from Telangana State of India.

Sample size: Adults of both sexes, total 500 nos.

Inclusion criteria: Medical and paramedical students between age group 18- 25 years.

Exclusion criteria: Teaching and non- teaching staff.

Questionnaire: The questionnaire was the key in collecting data for our study. It was designed for assessing the knowledge and attitudes of the target population towards consanguinity. Specific questions in the questionnaire addressed to the individual and his/her family for e.g– (1) any history of consanguinity in the family, (2) any genetic or hereditary disorder with/without the family history of consanguinity in an extended family of an individual, (3) opinion/suggestion regarding consanguinity, helped in knowing the current trends and prevalence of consanguinity. An extended family tree (3 generations w.r.t. WHO, 2010.)⁸ was prepared wherever required depending upon the complete history of the individuals considering the data from the questionnaire.

Lectures regarding consanguinity and its effects were given to different sample groups and feedback notes were collected. In offering preconception counseling for consanguinity, it is crucial to distinguish between families with a known genetic or inherited disorder and those with no such known disorder by taking a detailed family history and constructing a three generation family tree of the subjects.

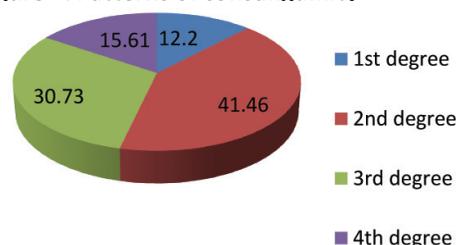
The doubtful cases were asked to attend the Prathima Institute of Medical Sciences and Hospital, or other regional health centres (of their wish) for further investigations, treatment and necessary follow up. In families with hearing, mental disabilities, informative family history along with clinical data and investigations can differentiate cases that are associated with consanguinity, or from cases caused by other factors.

RESULTS

In the present study, different variations are found between the consanguineous and non consanguineous groups which are as follows:

In our study, we found 205 consanguineous marriages out of the total target population of 500. The prevalence of consanguinity is found to be 41% against 51% Non-consanguineous marriages.

Figure 1: Patterns of consanguinity



The present study showed highest number of second degree relation/uncle-niece (41.46%) followed by third degree relation/first cousins(30.73%) and fourth degree relation/second cousins(15.61%) [Figure 1].

Table 1: Birth defects in offspring of consanguineous versus non-consanguineous parents

BIRTH DEFECTS IN OFFSPRING	Consanguineous marriage	Non-Consanguineous marriage	TOTAL
Yes	39 (19)	13 (4.4)	52
No	166 (81)	282 (95.6)	448
TOTAL	205	295	500

P=0.0000 (significant)

Here, we found 19% of consanguineous parents were having birth defects in their offspring while only 4.4% of non-consanguineous couples reported of birth defects and the difference was significant [Table 1].

Table 2: History of early death/still birth/miscarriage of siblings of target population

History of early death/still birth/miscarriage of siblings	Consanguineous marriage	Non-Consanguineous marriage	TOTAL
Yes	33 (16.1)	27 (9.2)	60
No	172 (83.9)	268 (90.8)	440
TOTAL	205	295	500

P=0.0187 (significant)

History of early death/ stillbirth/ miscarriage of siblings was found in 16.1% of consanguineous marriages and it is significant as compared to non-consanguineous marriage ie: 9.2%[Table 2].

When studied, Consanguineous versus non-consanguineous marriages with relation to different religions

showed that, out of total 455 Hindu population, 187(41.1%) were consanguineous against 268(58.9%) non consanguineous; out of total 19 muslims , 7(36.8%) were consanguineous; out of 26 Christians, 11(42.3%) were consanguineous. Here, $P=0.9250$ which is not significant.

While studying religions versus degree of Consanguinity we noted that 77 no. of Hindus (41.2%) preferred uncle niece union(second degree relationship), 4 Muslims (57.1%) favoured first cousins(third degree relationship). 6 Christians (54.5%) are also found to prefer second degree relationship.

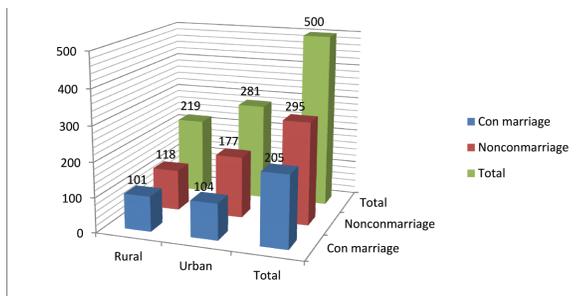


Fig 2: Consanguinity in Rural versus Urban population

While studying, Rural versus Urban prevalence , 46.1% (101/219) rural mass had overshoot 37% (104/281) urban population with regards to consanguinity. Here, $P=0.039$ (significant) [Figure 2].

In the target population, 184(41.7%) nuclear families and 21(35.6%) joint families were consanguineous. $P=0.368$ (not significant).

Table 3: Comparison of knowledge on consanguineous marriages in medical and paramedical students:

KNOWLEDGE	MEDICAL	PARAMEDICAL	TOTAL
Yes	343 (76.9)	19 (35.2)	362
No	103 (23.1)	35 (64.8)	138
TOTAL	446	54	500

$P<0.0000001$ (significant)

This table indicates that, medicos (76.9%) had more knowledge on consanguineous marriages in comparison to the paramedical students (35.2%) [Table 3].

When target population was asked about their opinion on cousin couples having children, it was answered as worrisome by 184(36.8%), unwise by 118(23.6%), normal by 115 (23%), understandable by 32(6.4%) and 51(10.2%) had given no response.Regarding health risk in consanguineous marriage in comparison to non consanguineous marriage, 460(92%) members found it higher, 20(4%) lower in consanguineous marriages, 11(2.2%) found it similar in both, while 9(1.8%) were not aware at all.

Out of total study population, 431(86.2%) had knowledge about the congenital deformities , 19(3.8%) about metabolic disorders, and 5(1%) about infectious diseases in the society. Unfortunately, 45(9%) were not aware of any health risks in consanguinity.In our study, 201(40.2%) opined it as unwise to marry a relative, 173(34.6%) as old fashioned, 63(12.6%) as normal, 40(8%) as understandable, while 23(4.6%) gave no response.

DISCUSSION

Marriage is one of the most vital & powerful bond of human relation. The family law in India takes into account the religious and cultural practices which are all equally recognized. One such familial-social bond is Consanguineous marriage. These marriages are favoured by different populations around the world bound to traditional customs, beliefs and to keep ancestral assets within the family⁹.

Around the globe consanguineous marriages are being practiced by many societies from time immemorial. In the Mahabharata, Arjuna and his son Abhimanyu both were said to have married their first cousins. Lord Buddha also married his first cousin¹⁰.

The Egyptian pharaohs performed akin marriages to keep wealth and power within the family to preserve the royal blood. They often married their own sister or half-sister; after a handful of generations, the offspring were mentally and physically unfit to rule. Cleopatra was born out of such a marriage¹.

Even the royal families of Europe practiced such marriages, because tradition did not allow them to marry people of non-royal class. The high amount of mentally retarded and handicapped royalties throughout European history shows the unhealthy consequences of this practice. It also predates the world's oldest monarchy, the Danish by 300years⁹. Charles Darwin who married his first cousin Emma Wedgwood, was one of the first experimentalist to demonstrate the untoward effects of inbreeding & consequences of consanguinity¹¹. Albert Einstein too married his second cousin, Elsa⁹.

In India, it is highly prevalent among Dravidian population called as Iroquois kinship¹². According to National Family Health Survey (1992-1993), consanguinity amounts to 16% of all marriages in India. However, this kinship varies from north (6 %) towards south (36%) of India with least in North-Eastern states^{13,14}. In North India, cousin marriage is proscribed and seen as incest for Hindus. Here, many communities do not allow candidates with common four surname/ Gotra to marry, which automatically rules out consanguinity⁹.

Levels of consanguinity varies from 4.5- 61.33% depending on factors like religion, caste and socio-economic factors. But generally inbreeding is most common in the more

traditional, rural communities. 30-40% of marriages in Andhra Pradesh (including Telangana) are cognate marriages¹⁵. One in two rural marriages in Andhra Pradesh & Tamil Nadu is consanguineous¹. In our study, we found prevalence of consanguinity to be 41%.

Prevalence of akin marriages in different parts of India is : Assam – 1.4%, Maharastra -26.4%, Pondicherry – 55%¹⁴; while the scenario in other parts of the world is –

Table 4 : Prevalence of consanguineous marriages in different parts of the world ^{5,9}

COUNTRY	PREVALENCE
North America, Western Europe	0.6%
Netherlands & United Kingdom	1-3%
Turkey	25-30%
Afghanistan	30-40%
Iraq	46.4%
Saudi Arab	67%
Pakistan	70%

Individual studies can give different risk figures dictated by various factors related to the study population.

The literature showed uncle – niece is commoner in Hindus & first cousin being preferred among Muslims ^{1,16,17}. Christian traditions discourage cousin marriage. But it is also location dependant among these communities¹⁶. While studying religions versus degree of Consanguinity we noted that 77 no. of Hindus (41.2%) preferred uncle niece union, 4 Muslims (57.1%) favored first cousins. 6 Christians (54.5%) are also found to prefer second degree relationship . A study in Mysore noted first cousin marriages (44.08%), uncle-niece marriages (46.81%) are equally significant in increasing congenital heart disease ¹⁸.

Many in the community are ignorant about the fact that consanguinity plays a negative role in their health. Although the situation appears better in the urban areas comparatively. While studying rural versus urban prevalence, 46.1% (101/219) rural mass had overshoot 37% (104/281) urban population with regards to consanguinity. Here, $P=0.039$ (significant). It correlates with the Pondicherry study which showed - 30.8% consanguinity (47.4%-first cousins, 23.4% uncle-niece) contributes considerably to infant mortality and morbidity with higher frequency in women from rural areas (Rural 35.6% vs urban 25.9% ; $p<0.001$) and among Hindus and Muslims than Christians¹⁹.

The birth defects and autosomal recessive/x-linked genetic disorders resulting from such marriages will continue to gain greater prominence in the overall spectrum of ill health²⁰. These cause a significant impact on world economy and productivity, and becomes a huge burden on the medical fraternity ⁹.

We found 19% of consanguineous parents having birth defects in their offspring, while 4.4% of non-consanguineous couples reported of birth defects and the difference was significant ($P < 0.0000$). The risk of birth defects is practically equal for all children with non consanguineous parents, irrespective of ethnic origin ²¹. But 5-10 times higher incidence of malformations is observed in consanguineous marriages²². A Guntur (Andhra Pradesh) based study among the babies with malformations, 3.8% babies were from consanguineous marriages while 15.1% were from Non Consanguineous marriages. Males were more affected as compared to females. When studied the type of consanguinity, second degree consanguineous was more²³. Congenital anomalies ($P= 0.02$) & low birth weight ($P=0.05$) were significantly higher among children with parental history of consanguineous marriage²⁴. The commonest being Neural tube defects ²⁵.

The progeny of first cousin marriages are not at an increased risk for birth defects in their children if they are married to a non relative²⁶. The frequency of malformations in different patterns of consanguineous marriages can vary in different places, based on marital habits, socio-economic status and environment of different places.

Consanguinity rate coupled by a large family size could induce the expression of autosomal recessive diseases (including very rare or new syndromes) in some communities⁵. In the target population, 41.7% nuclear families and 35.6% joint families were consanguineous. $P=0.368$ (not significant). In our study, history of early death/ stillbirth/ miscarriage of siblings was found in 16.1% of consanguineous marriages and it is significant ($P=0.0187$). Mortality in first cousin progeny is about 3-5% higher than in non-consanguineous offspring³. The risk of still birth doubles when parents are first cousins ²⁷. We found that, medicos (76.9%) had more knowledge on consanguineous marriages in comparison to the paramedical staff (35.2%). It is significant ($P<0.0000001$). Out of the total target population, 40.2% said marrying a relative as unwise, followed by 34.6% as old fashioned. Hence, education plays a key role in this aspect.

Cousin couples having children - was answered as worrisome by 184(36.8%), unwise by 118(23.6%), normal by 115 (23%), understandable by 32(6.4%) and 51(10.2%)

had given no response. Regarding health risk in consanguineous marriage in comparison to non-consanguineous marriage, 460(92%) members found it higher, 20(4%) lower in consanguineous marriages. 11(2.2%) found it similar in both, while 9(1.8%) were not aware at all. Out of total study population, 431(86.2%) knew about the congenital deformities in the society, 19(3.8%) about metabolic disorders, 5(1%) about infectious disease. Unfortunately, 45(9%) were not aware of any health risks in consanguinity.

As cognate marriages are culturally and socially favoured, we found 351(70.2%) members preferred this marriages for stronger family bonds. While only 37(7.4%) found it favorable for custom. Unfortunately 78(15.6%) gave no response. Our study revealed that, 418 (83.6%) members disagree with the continuation of consanguineous marriages in the society, while only 20(4%) agreed to the question. On the other hand, 62(12.4%) did not comment.

Regarding 'preferred provider of information', present study found that 234(46.8%) preferred gynecologist, 82(16.4%) government, 60(12%) community medicine provider, 56(11.2%) general practitioner, 5(1%) midwife, whereas 12(2.4%) did not respond. With respect to timing of offering information, maximum 457(91.4%) favoured it before marriage, 35(7%) said it to be before first pregnancy. Here, 4(0.8%) also preferred it during pregnancy.

Most of the subjects opined that awareness should be provided in a large scale by the Government in the form of audio-visual aids. Mass campaigning about the hazards of this kinship should be done at the community level, especially in the rural areas. Few also suggested that "adverse effects of consanguinity" should be included as a part of the study curriculum in higher secondary school level.

In our study we have found many relevant cases showing deleterious effects of consanguinity, carrier status as well as usefulness of having a non-consanguineous marriage.

Although there is some recognition of the genetic effects of consanguinity, these are greatly outweighed by social advantages such as greater family support and marital stability. Pre-marital negotiations regarding dowry are more easily conducted & less costly^{2,5}. Young age of consanguineous couples during marriage further complicates the situation⁹. It is sad that many movies in Tamil Nadu and Andhra Pradesh highlight and glorify consanguinity. Social benefits of consanguinity should not outweigh the biological damages¹.

Genetic counseling has a preventive, diagnostic, therapeutic and supportive value. It yields best results when given premaritally or at least prior to conception. Adopting better translational research concept and intervention

strategies will help these couples reach informed & intelligent reproductive decisions, with which they have to live throughout their lives. A research conducted in Andhra Pradesh on premarital genetic investigations and effects of genetic counseling, consanguinity was found in 86.04% cases while 73.25% presented family history of different genetic disorders²⁸.

Developing an understanding of these changes will require a wide ranging and multidisciplinary investigative approach for which community genetics is ideal to suite conditions in India. India needs to take a big leap in this aspect by setting up infrastructure to handle this grave situation.

CONCLUSION

Being local residents of Telangana State, we have witnessed the high prevalence of consanguineous marriages and its untoward effects. The social benefits of consanguinity should not outweigh the biological damages. Many in the community are ignorant about these facts. Improving socio-economic conditions e.g., improving female education, urbanization, smaller family size and a better access to health care will impact the effects of consanguinity, with a reduction in infant and child mortality in higher secondary school level. Public health programmes with multi-approach strategy, i.e, education about risk, pre-natal diagnosis, neonatal screening and Genetic counselling- yields best results if given premarital, or prior to conception. Our research work has helped to gain knowledge about the lifestyle, reasons behind akin marriages. It is expected to spread awareness and bring a change in the mindset of people regarding consanguinity & its effect. Hence, the present study can help people come to a conclusion about what is wrong in marrying a person whom they know since their childhood – The love that dare not speak its surname!

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