INTRODUCTION

Consanguinity is a term used to describe a wedlock between two individuals who are related by blood as a second cousin or closer, with the inbreeding coefficient (F) equal to or more than 0.0156. Here (F) is a measure of the proportion of loci at which a child can inherit identical gene copies from parents. It is a particular type of endogamy in which individuals with at least one common ancestor marry each other. Cousin marriage can be between double first cousins, first cousins, first cousins once removed, second cousins or third cousins who share at least one set of great-grandparents. Global prevalence of consanguinity and the patterns of different types of consanguineous marriages vary within and between populations according to ethnicity, culture, caste and religion. It is a genetic concept deeply...
ingrained in social norms of various societies living in the Middle East, Asia, and North Africa. Almost 6.5% of living couples in the world are in consanguineous unions and most of them are reported in Arab Muslim countries like UAE (54%) Qatar (54%), Saudi Arabia (42% - 67%), Iraq (47% to 60%), Sudan (44% - 63%), Jordan (64%), Kuwait (64%) and Iran (35-80%). 3 Almost one billion of the total world population still prefer to have intra-familial marriages as these unions are paramount to their kinship systems.1,4 A large population of these countries belong to younger age group and higher birth rate compare to world average.5

Close kin marriages were also practiced among Hindus, Jews, Buddhists, Christians, Parsis and Druze who live in southern and western regions of Asia.6 Historical literature corroborate such breeding practices in ancient Greece, Israel, classic Egypt and Orient regions of North and South Korea; where consanguineous marriages are now prohibited by law.7 In Ptolemaic Dynasty, the marriage practices were both incestuous and consanguineous as it was considered advantageous for a pharaoh to marry either his sister, half-sister or a cousin.8 Despite various cases of incestuous marriages in ancient Egypt, there is no authentic evidence of congenital anomaly or genetic diseases in their offspring.8

In Islam, intra-familial unions are not prohibited but no verse from Koran can be used as an evidence to support cousin marriages.9 In fact, a nuptial between real uncle and niece, marriage with real sister and wife of a son is strictly forbidden and considered incest like any other religion.2 However, there are some prominent examples in Islamic history like Prophet Muhammad who married his first paternal cousin Zainab Bint Jahsh and his daughter Fatima Zahra who married her father’s first cousin Ali. The second Caliph, Umar ibn al-Khattab, also married his first cousin, Atikah bint Zayd.

In England, the ratio of consanguineous marriages is less than 10% but the history of cousin unions in England has remained inconsistent and a topic of scholarly interest for many years.7 During the medieval period, first-cousin marriage was an anathema and taboo in the eyes of most people until 1540 when it was legalized.10 Cousin Marriage was popular among elite and middle-class British societies in the nineteenth century and almost one in every twenty marriages in landed gentry was between first cousins while in lower classes the rate was probably one in fifty.11

Despite the legality of cousin unions, such relationships remained under continuous scrutiny by medical researchers, biologists, social scientists and physicians. Close kin marriages were thought to cause deafness, blindness, insanity, infertility and countless other morbidities in the newborns.12 The injuriousness of consanguineous marriages was increasingly questioned in the late nineteenth century especially when Charles Darwin’s eldest son, George, a known mathematician, published an article suggesting restriction to cousin marriages by law. He propounded that the consequences of such practice might not be dreadful for an elite class but could be unsurmountable for the underprivileged community who cannot use quality health care services due to unaffordable treatment costs, if an anomalous child is born to them.11 Thereupon, no formal legislation was promulgated in Europe, but an apparent decline in cousin marriages to only 1% was observed in mid twentieth century.11

With expanding knowledge in social sciences and advanced research in medical genetics, queries about consanguineous marriages have become more complex. Couples, irrespective of their religion and cultural background seek preconception counseling and scientific answers to their questions mainly from the primary health care providers.1 In spite of being stigmatized and frowned upon by the modern society and medical researchers in Western world, cousin marriages (CM) are still practiced in many parts of the world.12

Nonetheless, the situation in the Islamic Republic of Pakistan has not changed to date and the overall prevalence ranges from 31.1 to 62%.14 Pakistan has one of the highest prevalence of consanguineous marriages in the world and remains stagnant.15,6 It is a multicultural country with diverse cultures, various castes and different Islamic sects living in its five distinct provinces. So far all the studies from different areas of Pakistan have shown a high number of consanguineous marriages and their association with various recessively inherited disorders. For instance, cousin marriages in Quetta are reported to be 31%, 46% in Lahore, 48.10% in Rawalpindi, 44.3 % in Jhelum, 48.50% in Gujrat, 52.1% in Faisalabad, 55% in Peshawar, and 66 % in Malakand district.16 However, there is no formal study from the capital city, Islamabad, which in many ways is a microcosm of Pakistan. The aim of this research was to determine the prevalence of consanguineous marriages, its association with the sociodemographic characteristics, to ascertain reasons for preference of cousin unions and to determine if there is a need for pre-natal genetic counseling.

METHODS

Study Design

This was a cross-sectional survey conducted for three months at 125 bedded Maternal and Child Health Center (MCH) of a tertiary care hospital. A sample size was calculated by applying following epidemiological formula.

\[ n = \frac{[1.96^2 \times P(1-P)]}{\varepsilon^2} \]

Where n is number of person to be interviewed or sample size, P is the recent percentage of the population with the same characteristics (cousin unions) determined through
previous research (50%) and \( \epsilon \) is the standard error taken as 6%.

\[
n = \frac{(1.96^2 \times 50 \times 50)}{6^2} = \frac{9600}{36} = 267
\]

Expecting a 20% refusal rate, the sample size approximated to 300 respondents. Data were collected by using a pre-validated questionnaire to interview women selected through convenience sampling. The questionnaire was initially developed into the English language; however, to make it more comprehensive for the participants who cannot speak or understand English, it was translated into the Urdu language. It mainly comprised of closed-end questions with one open-ended query of a reason for preference or refutation for such union. At first, 30 women were interviewed for a pilot study and the external validity and reliability was confirmed to proceed with further interviews. None of the approached women refused to participate in the study, and the response rate was 100%.

Independent variables were age, age at marriage, residential area, educational status, province of origin, degree of consanguinity, caste, the number of children, miscarriages and number of anomalous children. Participants belonged to different castes like Syed; considered as the most honorable and highest among all other castes in Pakistan, then Rajput, Chaudry, Awan, Raja, and others. Women who were married to the distant relatives or within clans were taken as non-cousin unions. All married women within the age range of 15-49 years, living with husbands and presenting to the obstetric and gynecological clinic were included. Single, Divorced, Widow, infertile, menopausal and laboring women were excluded from the study.

**Data Analysis**

Data were entered and analyzed with Statistical package for social sciences SPSS 21. Descriptive analysis was performed, and frequencies were calculated. Chi-square test was applied to find an association between two categorical variables and a \( p \) value of <0.05 was taken as statistically significant.

**Ethical Considerations**

Ethical approval for the study was obtained from the Ethical review committee. An informed verbal consent was taken from participants who were free from coercion to answer the questions and were allowed to leave the study at any point. Personal information like name and residential address was safeguarded by locking questionnaires into cabinets to maintain anonymity and confidentiality. The statistical tool used for analysis was password protected and accessible only by the researcher.

**RESULTS**

**Sample Characteristics**

The mean age of women was 28.3 \( \pm \) 5.82 (SD) years. The mean age of the women in consanguineous union was 21 years as compared to 24 years age of women in non-consanguineous union. Mean age at marriage of all women was 22.5 years \( \pm \) 4.27 (SD). There was no significant difference in the mean age at marriage in cousin union (23.3\( \pm \) 4.1 SD) and the mean age at marriage in non-consanguineous union (21.2 years \( \pm \) 4.1 SD).

The minimum age of woman at marriage was 13 years and maximum was 37 years. At the time of study approximately 38\% (n=114) of women were nulliparous, 25.7\% (n=77) had only one child, 15\% (45) had two children and remaining had more than 2 children with maximum 10 children to only one woman. Consanguineous couples had more children with a mean of 3 children as compared to mean of 2 children born to a non-consanguineous couple. Table 1 shows the sociodemographic characteristics of the participants.

<table>
<thead>
<tr>
<th>Socio-demographic characteristics</th>
<th>Consanguineous union</th>
<th>Non-consanguineous union</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mean age (years)</td>
<td>21</td>
<td>24</td>
</tr>
<tr>
<td>Mean age at marriage (years)</td>
<td>23.3</td>
<td>21.2</td>
</tr>
<tr>
<td>Mean number of children</td>
<td>3</td>
<td>2</td>
</tr>
<tr>
<td>Number of anomalous babies</td>
<td>Nil</td>
<td>2</td>
</tr>
<tr>
<td>History of miscarriage</td>
<td>32%</td>
<td>30%</td>
</tr>
<tr>
<td>Residence in urban areas</td>
<td>44.3%</td>
<td>70.8%</td>
</tr>
<tr>
<td>Residence in rural areas</td>
<td>55.7%</td>
<td>29.2%</td>
</tr>
<tr>
<td>Overall education</td>
<td>82%</td>
<td>94.6%</td>
</tr>
</tbody>
</table>

Marriages were broadly categorized into consanguineous and non-consanguineous for data interpretation. Out of 300 participants, 62\% (n=187) had cousin marriages, and 38\% (n=113) had non-cousin marriages. Overall first
cousin marriage predominated with 61.5% (115/187) followed by 20.5% (38/187) second and 18% (34/187) third cousin nuptials. Matrilineral marriages were 64% among first cousin marriages and 36% were patrilineral Figure 1.

The majority of participants belonged to the province of Punjab (84%), a few were from Khyber Pakhtunkhwa (13%) and Sindh (3%); while none of the participants were from Baluchistan. Some prominent castes of women in consanguineous marriages were Rajput (n=35, 18.7%), Awan (n=28, 14.9%), Syed (n=21, 11.2%) and Chaudry (n=14, 7.5%) and 47.6% (n= 112/187) of the participants in consanguineous unions belonged to various other castes like Sheikh, Musalli, Jutt, Gujjar, Qureshi, Pathan, Ghumman and Malik. Overall 63% (n=190) of women were aware of a general notion that consanguineous couples have double risk to have babies with congenital anomalies or recurrent miscarriages. They thought that couples should receive pre-genetic and preconception counseling by the doctors, health care providers or through social media programs on television, radio and social websites.

Among consanguineous marriages, 82% (152/187) of women had attended 5 or more years of schooling while 18% (33/187) were illiterate. Out of 82 % (152/187) of educated cousin unions, 7% (n=13) had primary school education, 24.5% (n=32) had secondary school education, 17.2% (n=32) had higher secondary education, 25.1% (n=47) had Bachelor’s degree and 8% (n= 15) had Master’s degree as shown in Figure 2.

Approximately, 94.6% (107/113) of non-cousin unions were educated, and approximately 65% of women had Bachelors or higher degree while 5.4% (6/113) illiterate.

The overall literacy rate was greater among non-consanguineous union in contrast to consanguineous unions. There was a significant association between educational status and preference for consanguineous marriage ($x^2=131.24$, df=5, $p=0.0001$). Women with higher education level were against cousin marriages while those with less education showed positive attitudinal behavior towards cousin unions.

On the whole 54% of women were living in urban areas and 46% were residing in rural areas. Among women in non-consanguineous nuptial, 44.3% (50/113) were living in urban areas while 55.7% (63/113) were living in rural areas.

Conversely, 70.8% (132/187) of women married to cousins were living in rural areas and 29.2% (55/187) were living in urban regions. Bivariate analysis showed a significant relationship between place of residence and preference of marriage within the family ($x^2=7.13$, df= 1, $p = 0.0001$). Women hailing from rural areas were in strong favor of consanguineous marriages unlike women who were living in urban cities.

No anomalous child was reported by any women married to a close relative, but two anomalous babies (n=2) were confirmed in the non-consanguineous group. In the consanguineous group, 32% of women had a previous history of miscarriages and similarly 30% in the non-consanguineous group. No significant association was found between the number of miscarriages and type of marriage ($x^2=3.00$, df=5, $p=0.69$). None of the women ever had prenatal genetic counseling in any pregnancy while all (100%) were keen to have it for their future pregnancies.

**Reasons for Preference of Intra-familial marriages**

Majority women in consanguineous union (90%) revealed that they had no liberty or say to choose their
spouse and their parents were the prime decision makers of their marriage. Many of these women (96.7%) still favor such marriages for their progeny due to family traditions and a sense of security for daughters. In their opinion, they get better treatment by their spouses when chosen within the family and the relationship is thought to be more stable. Women of Syed caste strongly favored cousin marriages in a view to maintain the purity of their lineage from Prophet PBUH descendants.

Table 2: Reasons of preference for cousin marriages among women who were in consanguineous union.

<table>
<thead>
<tr>
<th>Reasons of preference (n=187)</th>
<th>Yes (n)</th>
<th>No (n)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Security</td>
<td>181</td>
<td>6</td>
</tr>
<tr>
<td>Property/Land</td>
<td>100</td>
<td>87</td>
</tr>
<tr>
<td>Traditions/customs</td>
<td>181</td>
<td>6</td>
</tr>
<tr>
<td>Religious beliefs</td>
<td>50</td>
<td>137</td>
</tr>
<tr>
<td>Others</td>
<td>20</td>
<td>167</td>
</tr>
</tbody>
</table>

On the other hand, 53.4% (100/187) thought that such unions allow preservation of lands and wealth within families. Only 26.7% (50/187) stated that such unions are preferred on religious grounds. A few women in non-consanguineous union (22/113) stated that a cousin marriage strengthens the relationship between husband and wife especially of those who live in a conservative milieu. Religion was considered the least driving factor and more of a sociocultural reason for marrying within families. Reasons to prefer consanguineous marriages are summarized in Table 2.

DISCUSSION

Our present study has substantiated previous figures of 62% of consanguineous marriages in Pakistan. Various studies conducted in 1993, 1996 and 1998 have also shown similar prevalence of 60% cousin unions in Pakistan. It was observed that more than half women in cousin union still prefer cousin marriages for their future generations.

Pakistan is a large power distance country with a predominantly patriarchal society. Respect of parents and elder siblings or brothers is a lifelong integrity who, in majority cases, are the sole decision makers for marriages. Most of the time they choose mates from the families and preferably from father’s side to avoid wealth sharing out of a family, for financial reasons like dowry and to maintain family traditions. We observed the same patterns among our respondents who showed a strong inclination towards cousin marriages for their progeny.

Several reasons cited in the literature are the ease of pre-marriage negotiations about property and family structure or monetary benefits in the context of dowry and bride wealth payments. Similarly, not many respondents in the present study quoted religion as the main factor for cousin marriage but explained that such unions are still ideal due for family bond strengthening, for transmission of cultural values and retention of wealth and property within same family.

Results from the present study also illustrated a significant relationship between education, residence, and preference to marry a cousin. These findings are again in accord with results of Pakistan Demographic Health Survey (PDHS) 2012-13 which showed that 54% of all consanguineous marriages are in rural areas and 38% are from urban areas. The association of consanguinity with low socioeconomic status, younger age at marriage, residence in rural areas and less education has already been studied among significant populations of different cities in Pakistan and the present study verifies this association. Past studies from Japan, Turkey, Pakistan, Iran, and Yemen, have also highlighted the inverse relationship between education and consanguinity. It has been reiterated that higher education reduces the rate of cousin marriages and this fact has been anchored by aforementioned results that conduct to prove that education and residence in an urban region improves the autonomy and understanding of a woman towards her health needs since majority women with higher education had non-consanguineous marriage and did not prefer cousin marriages.

No significant relationship was observed between consanguinity and anomalous children or consanguinity and number of miscarriages or child mortality unlike some regional studies that explained an association between consanguineous marriage and child mortality. The most plausible explanation to these results is the non-probability sampling technique that can lead to sampling bias and the results cannot be generalized to the entire population. However, there are many studies that failed to establish any positive relationship between fetal loss and consanguinity. Also, while drawing conclusions on association between cousin marriages and birth defects, one should not ignore young maternal age, maternal education, rural residence and poor nutrition as confounding factors.

Despite increasing educational level in Pakistan, the prevalence of cousin marriages remain unchanged. This issue in Pakistan deserves special attention as the incidence has risen from 38.8% in 1978 and 1979 to 61.2% at the present time, and this sharply increasing practice can endanger community health and economic development of this region. A suggestion by various scientists to legally ban cousin marriages in societies where it is strongly supported on cultural ground; is not only difficult and controversial, but unthinkable. In such circumstances prenatal genetic counseling is an indispensable tool.

Today couples, everywhere in the world, are more receptive and often seek prenatal genetic counseling and prenatal diagnosis in the context of many genetic diseases.
like thalassemia, Down’s syndrome, induced abortions on medical grounds and many others. Therefore, there is a need to recognize consanguinity as a public health issue that needs pertinent global attention, resource mobilization, and advocacy.

Admittedly, present study has few caveats worth mentioning. Firstly, due to non-availability of funds, absence of registries and computerized programs, participants were recruited by using convenience sampling technique. Hence the results of this study cannot be generalized to the whole population. Secondly, the positionality of a researcher as an insider could have affected the response of participants leading to research bias.

CONCLUSION

Uncountable studies had been conducted in the past to evaluate social trends towards consanguineous marriages in Pakistan; and to date, no change has been observed. The results obtained from this study calls for national awareness programs and prenatal genetic counseling in antenatal clinics or on community level to avoid the repercussions of consanguineous marriages on future generations. There is further need for a broad, in-depth survey on a national level to assess general attitude and perceptions of Pakistani population towards prenatal genetic counseling and consanguinity.

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Ethical approval: The study was approved by the Institutional Ethics Committee

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