Awareness Regarding Risks of Genetic Disorders Due To Consanguineous Marriages

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ABSTRACT:- Consanguinity - blood relation, descended from the same ancestor, is a deeply rooted social trend among one-fifth of the world population. On average, first cousins have an extra risk of 1.7–2.8% of having a child with many genetic disorders. The current study was conducted to evaluate the awareness regarding consanguineous marriages in laymen population and medical personnel, rate of consanguineous marriages and the risks of complications, infant deaths and genetic disorders due to it. It is a survey based study carried on medical professionals (N=100) and laymen population (N=200) including literate (N=100) and illiterate (N=100), age 20-50 from different areas of Karachi. The answers were recorded as open and close ended. From our data we found that 53% of medical population is familiar with the term consanguineous marriages, whereas there was no awareness in illiterate laymen population regarding this term. 86% medical professionals and 46% of literate population agreed that consanguineous marriages can be one of the reasons of genetic disorders for offspring while 76% of laymen illiterate population disagreed over this statement. 74% illiterate, 51% literate and 40% of medical population are involved in consanguineous marriages and the families of 31% medical professional, 33% literate laymen and 46% illiterate population, also suffer from early infant death and birth defects. 77% medical professionals, 33% literate people and 14% illiterate people are in favor of proper medical checkup and testing before consanguineous marriages. Consanguineous marriages are favored in our society that leads to high risk of genetic disorders, infant deaths and complications at time of giving birth. Awareness needs to be created about issues regarding consanguineous marriages and need to visit genetic counselor for proper medical checkups and testing.

Key Words: consanguineous, genetic counselor, genetic disorders, offspring

I. INTRODUCTION

Consanguineous marriage refers to marriage between close relatives which leads to increase risk of genetic disorders and still births. [1] It is a global practice which has been in existence since early civilization of humans. Its occurrence varies worldwide with respect to culture, religion and geographical location. [2] About one third of the human population of the world lives in countries with a preference for consanguineous marriage and at least 8.5% of children have consanguineous parents. [3]

Marriage between couples who share at least one common ancestor is defined as consanguinity or inbreeding. [4] Consanguinity has received a great deal of attention as a potential risk factor for many adverse health outcomes, because it favors the reemergence of recessive deleterious alleles that run in families. Stillbirths and spontaneous abortions, as well as infant death, have been shown to be positively correlated with consanguinity. Consanguinity was also identified as a risk factor for several morbid conditions including congenital abnormalities and other birth defects, mental retardation, deafness, β-thalassemia, chronic renal failure and neonatal diabetes mellitus [5]

Consanguineous unions afford the possibility that susceptibility genes identical by descent may be inherited through the relatedness of child-bearing couples. [6] Inbreeding increases the chances of the expression of deleterious recessive alleles by increasing homozygosity and therefore has the potential to decrease the fitness of the offspring. With continuous inbreeding, homozygosity is increased and variation in genes is lost which causes recessive deleterious alleles to be expressed in homozygotes. By pairing chromosomes of similar genomes, the chance for these recessive alleles to pair and become homozygous greatly increases, leading to offspring with autosomal recessive disorders. [7]

In many populations there is a strong preference for consanguineous unions, most commonly among first cousins, and marriage outside the family is considered as a risk and disruptive option. [8] Recent survey by Pakistan Demographic and Health (DHS) show that two-thirds of marriages in Pakistan are consanguineous. This survey examined the relative importance of the three commonly perceived reasons for such marriages: religious, economic and cultural. The major reasons for a preference for consanguineous marriages are sociocultural rather than any perceived economic benefits. [9] Cultures with preferences for consanguineous unions believe that these unions are the best opportunity for a compatible marriage, since both parties are well
aware of each other’s lifestyle. It is thought that the bride is accepted more readily and undergoes less scrutiny in such marriages. [10]

It is commonly believed that Islam favors marriage between close relatives other than the proscribed ones such as those between siblings, parent and child, uncle and niece or aunt and nephew. However, no passage in the Koran can be interpreted as encouraging consanguineous marriages. The only clan groups in Pakistan that practice endogamy for religious reasons are Syeds (who claim to have lineage associated directly with offspring of the Prophet) and Qureshis (who claim to have lineage associated with the patrilineal of the Prophet), and therefore are concerned with keeping the lineage ‘pure’. [11] Within the major religions there are quite marked differences in attitude to close kin marriage. Thus in Christianity, the Orthodox churches prohibit consanguineous marriage, the Roman Catholic church currently requires Diocesan permission for marriages between first cousins, and the Protestant denominations permit marriages up to and including first cousin unions.

A similar degree of non-uniformity exists in Hinduism. The Aryan Hindus of northern India prohibit marriage between biological kin. By comparison, Dravidian Hindus of South India strongly favor marriage between first cousins. [12-13] The consolidation of family property has been one of the other commonly cited economic reasons for continuation of cousin marriages in the Middle East and South Asia in general. Dowry or jehez is therefore the major economic transaction that underpins marriage decision-making. It is generally true that the size of the dowry is a consideration in opting for cousin marriages. [14-15]

According to study by DrShaneelaAsad, a pediatrician at a leading hospital in Karachi, There is a 25% chance that an offspring of a cousin marriage will contract a disease or disorder coming down from one parent. Doctors cite deafness, blindness, mutism, asthma, Down’s syndrome, thalassaemia and other skin, heart, kidney and neuro-degenerative conditions as consequences of inter-breeding between close relatives. [16]

Genetic counseling needs to be promoted. A genetic counselor should focus on being sensitive in understanding and handling the issues being brought to him. They should avoid stereotyping. For enhancing services the counselor should be multilingual or an interpreter should be present [17-18]

The present study has been designed to evaluate the trend of cousin marriages in Medical and Non medical population, to check the knowledge and attitude of different populations that cousin marriages can increase risk of genetic diseases and to observe concept and awareness regarding genetic counselor or checkup before cousin marriages.

II. METHODOLOGY

It is a randomized survey based study carried out on N=300 comprising of Medical population (N=100) and 100 each Literate and Illiterate population. The data was collected from different locations in Karachi including universities and Food outlets where as for medical population data was collected from different Clinics as well as Private and Government Hospital. The survey included both sexes (male and females) and age limit was from 20-50 years. Data was collected from February –April 2016. The answers were recorded both as open and close ended. For Illiterate population the questions were explained to laymen in urdu (native language) and the answers were then marked. Consent was taken from the individuals prior to the study.

2.1 Inclusion Criteria:
Medical Population included Medical students as well as medical officers, residents and consultants. Married and unmarried both population were considered but data was not interpreted based on this.

III. RESULT

The results were evaluated using SPSS Version 20. Percentages have been presented in Table 1 where as Two-way Anova followed by Post hoc Tukeys test for evaluation of significance among groups is presented in Table 2

<table>
<thead>
<tr>
<th>Table 1: Percentage Evaluation of Populations Regarding Consanguineous Marriage Awareness</th>
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<tr>
<td>Awareness regarding term Consanguinity</td>
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<td>Yes</td>
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<td>Awareness that Consanguineous marriages increase risk of genetic disorder</td>
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<td>Trend of consanguineous marriage in family</td>
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**Awareness Regarding Risks of Genetic Disorders Due To Consanguineous Marriages**

| Prefer Treatment before consanguineous marriage | Yes | 77% | 33% | 14% |
| Awareness regarding role of Genetic counselor | Yes | 74% | 24% | 9% |
| Counseled by Doctor against repeated consanguineous marriages in family | Yes | 71% | 47% | 19% |

Table 2: Comparative Evaluation of Medical population with different Laymen populations regarding awareness of Consanguineous Marriage:

<table>
<thead>
<tr>
<th>Table 2: Comparative Evaluation of Medical population with different Laymen populations regarding awareness of Consanguineous Marriage:</th>
<th>Literate Laymen</th>
<th>Illiterate Laymen</th>
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<tr>
<td>1 Awareness regarding term Consanguinity</td>
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<td>3 Trend of consanguineous marriage in family</td>
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<td>4 Trend of Birth Disorders or Early Infant deaths in family</td>
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SPSS Version 20 has been used. Two way Anova applied. Post hoc analysis by Tukeys test shows p values as P<0.05 significant * P<0.01 moderately significant ** P<0.001 highly significant *** IS insignificant

**IV. DISCUSSION**

The practice of consanguineous marriages is also related to dowry system to strengthen the relationship between the bride and groom families and it also lowers the cost of dowry. [29] We have surveyed about awareness regarding risk of genetic disorder due to consanguineous marriages from 300 individuals including medical professionals, literate and illiterate laymen, both male and female living permanently in Karachi.

Table 1 shows the familiarity of the term “consanguinity”. Our result shows 53% of medical professionals are familiar of the term Consanguinity whereas only 23% literate laymen have heard this term and 0% of illiterate people are aware of this term. It also shows that 86% medical professionals agree that consanguineous marriages can increase risk of genetic disorders, 24% of Illiterate people whereas 46% of literate people agreed with it.

Literature study carried out in Dubai and Al-ain comprising of N=2200 aged > 15 years showed that the rate of consanguineous marriage was 50.5% and parity, gravidity, ages and number of children were similar in consanguineous and non-consanguineous groups. However children born to consanguineous unions also had significantly higher incidences of illnesses (37%) than those of non-consanguineous unions (29%). The occurrence of malignancies, congenital abnormalities, mental retardation and physical handicap was significantly higher in offspring of consanguineous than non-consanguineous marriages. [19-20]

Table 1 also shows that 40% of the medical related families have cousin marriages, 74% illiterate people whereas 51% of literate people are also engaged in consanguineous marriages. Literature study conducted in the Department of Medical Genetics in Rabat on 176 families with autosomal recessive diseases diagnosed and confirmed by clinical, radiological, enzymatic or molecular investigations showed consanguineous marriages comprised 59.09% of all marriages. The prevalence of consanguinity in Morocco was found to be 15.25%. [21] Another literature study showed 32.5% couples observed cousin marriages in Iran. [22] Another literature evaluation showed 5401 marriages were evaluated from years 1950 to after 1980. The study divided the marriages in 3 generations and showed that first cousin marriages were 20.2% in years before 1950; it was 28.5% between 1950-1979 and 19.5% after 1980. [23-24]

Table 1 also shows that 31% of medical professionals, 46% of illiterate people and 33% of literate people have observed early infants deaths and birth disorders in their families as a result of consanguinity. Marriage between first cousins doubles the risk of children being born with birth defects, according to a study seeking answers to the higher than expected rates of deaths and congenital abnormalities in the babies of the Pakistani community. The offspring of consanguineous unions may be at increased risk for recessive disorders because of the expression of autosomal recessive gene mutations inherited from a common ancestor. [25-26]
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Data presented in Table 1 shows 77% of medical professionals, 14% of illiterate people and 33% of literate people stated that they will prefer the treatment before consanguineous marriage, which mean 86% illiterate people and 67% literate people are still not aware of the risk of consanguinity that may lead to stillbirths and infant deaths to their families. Tests that will contribute to lessen the risk of consanguinity defects would include blood grouping and Rh factor, screening for thalassemia, sickle cell anemia, hepatatitis, sexually transmitted diseases like syphilis and AIDS. These tests should be correlated with past medical history and family history. Genetical analysis for inherited genetic variants is performed for several purposes: diagnosis of individuals with symptoms, determination of future disease risks in asymptomatic individuals. Maternal serum screening and high resolution fetal ultrasound at 20-22 weeks can also be done. Education of the public in general and of primary health personnel in particular is an important pillar in clarifying the health and social effects of consanguineous marriages. [27-28]

Table 1 also shows that 74% of the medical personnel are aware about role of genetic counselor, 24% literate laymen whereas 9% of the illiterate people were surprisingly aware of it. Genetic counseling yields best results when done premaritally or at least prior to conception. A non-judgmental attitude towards consanguineous couples is essential on the part of the counselor, to establish good communication channels and to foster effective working relationships between the medical profession and communities where consanguineous marriages are prevalent. [29-30]

It has been presented in Table 1 that 71% of the medical professional agreed that they were counseled by doctors before consanguineous marriage similarly 19% Illiterate people and 47% literate people agreed that doctors counseled them to marry outside family. One of Dubai’s leading geneticists, Dr Mahmoud Taleb Al Ali, who is also an Emirati, readily admits it would be a waste of resources to try to discourage interfamily marriages. According to him "People are still marrying their cousins even though you tell them the science. The important thing is they come forward when they want to marry, especially if they have genetic diseases, there are ways to help them.” [31-34]

Table 2 shows there is highly significant awareness regarding term consanguineous marriage, it being one of the major causes of genetic diseases, awareness regarding genetic counselor etc in medical population as compared to laymen (Both literate and Illiterate)

V. CONCLUSION

From our study we have concluded that the awareness regarding complications of consanguineous marriages in laymen population is not enough. Awareness needs to be created on issues regarding consanguineous marriages and increase risk on adverse health outcome of infants. It’s mandatory to make the population accept the reality that complication of child birth can be due to consanguineous marriages and they should visit genetic counselor for proper medical checkups and testing.

REFERENCES

Awareness Regarding Risks of Genetic Disorders Due To Consanguineous Marriages


