ASSESSMENT OF CONGENITAL MALFORMATIONS IN CONSANGUINEOUS MARRIAGES AMONG THE FAMILIES OF STUDENTS OF SAIDU MEDICAL COLLEGE SAIDU SHARIF.

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ABSTRACT

Consanguineous marriage is customary in many societies, which leads to an increased birth prevalence of infants with severe recessive disorders (congenital malformations). All these disorders play a significant role on world economy, productivity and become a huge burden on medical fraternity. Majority of these disorders are preventable through proper “Premarital consultation / counseling” of the male and female before wedding for checking of “Marital compatibility”.

Objective of this observational study is to assess the prevalence of congenital malformations in consanguineous marriages among the families’ of students of Saidu Medical College.

Material & Methods:

Duration of study was 3 months.

Sample size Sampling Techniques: Stratified Randomized technique was used for selection of 100 houses/families of students of the college, with equal strata taken from first to final year MBBS classes.

Result: From 504, 280(55.56%) of the children were from consanguineous marriages, while 224(44.45%) were from non consanguineous marriages. From 25 cases with congenital anomalies, 23 (8.2%) cases were from consanguineous marriages while only 2 (0.9%) cases were from non-consanguineous marriages. Our study shows that there is a significant correlation between parental marriages and the prevalence of anomaly. 54 % couples out of 100 families were belonged to consanguineous marriages, of which 24 %, 17% and 13% marriages were belonged to 1° cousin, 2° cousin and other cousins respectively. 46 % out of 100 families were belonged to non-consanguineous marriages.

KEY WORDS: Congenital malformations; recessive disorders; consanguineous marriage

INTRODUCTION

It is estimated that one billion of the current global population live in communities with a preference for consanguineous marriage¹. Around the globe consanguineous marriages have been practiced by many societies from time immemorial⁵. Consanguinity is widely practiced in several global communities with variable rates depending on religion, culture, and geography. It is widely practiced in Asia, North Africa, Switzerland, Middle East, some parts of China, Japan and fishermen communities in Europe and America⁶. Generally, the highest rates of marriages to close relatives are consistently reported in the more traditional rural areas and among the poorest and least educated in society⁷. Globally, the most common form of consanguineous union contracted is between first cousins, in which the spouses’ share 1/8 of their genes inherited from a common ancestor.

The risk of birth defects in first-cousin marriages may be estimated to be 2 to 2.5 times the general population rate, mainly due to the expression of autosomal recessive disorders⁸. Frequency of consanguineous marriages was higher among parents of offspring with congenital malformations compared with the figures for the
general population in all studies reported. Consanguinity without known genetic disease in the family appears to cause an increase in mortality and malformation rate which is extremely marked in the children of incestuous mating, but which is of little significance when the relationship is more distant than that of first cousins. Consanguineous marriages are generally thought to be more stable than marriages between non-relatives due to various reasons.

Arab populations have a long tradition of consanguinity due to socio-cultural factors. Many Arab countries display some of the highest rates of consanguineous marriages in the world ranging around 20-50% of all marriages, and specifically favoring first cousin marriages with average rates of about 20-30%. Socio-cultural factors, such as maintenance of family structure and property, ease of marital arrangements, better relations with in-laws, and financial advantages relating to dowry seem to play a crucial role in the preference of consanguinity in Arab populations. Consanguinity in the Arab World is not only confined to Muslim communities. Several other communities, including the Lebanese, Jordanian, and Palestinian Christian populations, have also practiced consanguinity, but to a lesser extent than Muslims. In South East Asia the prevalence of consanguineous marriages is ranging from 20-70%. One in two rural marriages in Tamil Nadu and Andhra Pradesh is consanguineous.

Pakistan is a country where consanguineous marriages are a social norm and are widely practiced. In Pakistan consanguinity was seen in 60% of marriages out of which 80% were between first cousins. Considering the population of Pakistan and high fertility rate with the cultural background of consanguineous marriages, the problems of inherited disorders and congenital malformations may be significant. All these disorders play a significant role on world economy, productivity and become a huge burden on medical fraternity. Strengthening family relationships and keeping wealth in family are some of the reasons of consanguineous marriages in Pakistan.

We performed this observational study in order to obtain the frequency of consanguineous marriages in our society and to evaluate the consequences of such marriages related to congenital malformations in offspring and to propose appropriate procedures for awareness of people of the futurity.

MATERIAL AND METHODS

Setting: This study was conducted in Saidu Medical College Saidu Sharif from 1st October to 30th December 2012.

Population:
All the families of the MBBS students of Saidu Medical College Saidu Sharif, Swat.

Sample size
100 families of the MBBS students of Saidu Medical College Saidu Sharif, Swat were selected as a sample out 406 total families. They have total number of 504 children, 292 male and 210 female children. The numbers of consanguineous marriages were 54, having 280 numbers of children. Out of theses 280 children, 158 were male while 122 were female. The numbers of non-consanguineous marriages were 46, having 224 numbers of children, out of which 134 were male while 90 were female children.

Sample Technique
The whole population was divided into five strata. One stratum of 20 families selected from each MBBS class. They were randomly selected from each class, from first to final year MBBS.

Inclusion criteria: All willing families of the MBBS students of Saidu Medical College Saidu Sharif, Swat were included. All the couples having children are included.

Exclusion criteria: Non-willing couples and (sterile) couples without children.

Data collection tool
A pretested questionnaire was used to collect data. Data was analyzed by ordinary calculator and presented in appropriate table.
Data collection procedure

Data was collected through pre-tested questionnaire with the help of volunteer MBBS students of Saidu Medical College, Saidu Sharif, Swat after the consent of the informant. The result was presented in tabulated form and percentage of prevalence of consanguineous marriages and percentage of prevalence of congenital malformations in their children is shown in Table 1, 2 & 3.

Statistical analysis: all collected data was analyzed by ordinary calculator

<table>
<thead>
<tr>
<th>Numbers of Marriages</th>
<th>Type of Marriage</th>
<th>Consanguineous Marriages</th>
<th>Non-Consanguineous Marriages</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>First cousin</td>
<td>Second cousin</td>
<td>Others</td>
</tr>
<tr>
<td>Total</td>
<td>24</td>
<td>17</td>
<td>13</td>
</tr>
<tr>
<td>Percentages</td>
<td>24%</td>
<td>17%</td>
<td>13%</td>
</tr>
</tbody>
</table>

TABLE 2
STATUS OF ABNORMAL CHILDREN IN FAMILIES OF VARIOUS TYPES OF MARRIAGES

<table>
<thead>
<tr>
<th>Numbers of children</th>
<th>Type of Marriage</th>
<th>Consanguineous Marriages</th>
<th>Non-Consanguineous Marriages</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>First cousin</td>
<td>Second cousin</td>
<td>Others</td>
</tr>
<tr>
<td>Total</td>
<td>101</td>
<td>101</td>
<td>78</td>
</tr>
<tr>
<td>Normal</td>
<td>90</td>
<td>92</td>
<td>75</td>
</tr>
<tr>
<td>Abnormal</td>
<td>11</td>
<td>9</td>
<td>3</td>
</tr>
<tr>
<td>Percentage of abnormal children</td>
<td>3.9</td>
<td>3.2</td>
<td>1.1%</td>
</tr>
</tbody>
</table>

TABLE 3
STATUS OF SYSTEM WISE CONGENITAL MALFORMATIONS IN CHILDREN

<table>
<thead>
<tr>
<th>Malformations system</th>
<th>Frequency According to the Type of Marriage</th>
<th>Consanguineous Marriages</th>
<th>Non-Consanguineous Marriages</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>First cousin</td>
<td>Second cousin</td>
<td>Others</td>
</tr>
<tr>
<td>Nervous system</td>
<td>5</td>
<td>6</td>
<td>1</td>
</tr>
<tr>
<td>(mental retardation, speech, dumpy)</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Cleft Lip</td>
<td>1</td>
<td>0</td>
<td>2</td>
</tr>
<tr>
<td>Cleft Palate</td>
<td>1</td>
<td>0</td>
<td>2</td>
</tr>
<tr>
<td>Congenital Heart Disease</td>
<td>2</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Ear (Deafness)</td>
<td>1</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Muscular (cerebral)</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Cleft Anus (Aurea)</td>
<td>0</td>
<td>1</td>
<td>0</td>
</tr>
</tbody>
</table>

RESULTS

From 504, 280 (55.56%) of the children were from consanguineous marriages, while 224 (44.45%) were from non-consanguineous marriages, having 25 children with congenital anomalies. In couples having consanguineous marriages, the total numbers of children were 280, 158 male and 122 female. From 25 cases with congenital anomalies, 23 (8.2%) were from consanguineous marriages while 257 (91.8%) were found normal. Two children (male) were suffered from severe congenital cardiac abnormalities and found dead at the time of birth.

54% couples out of 100 families were belonged to consanguineous marriages, of which 24%, 17% and 13% marriages were belonged to 1st cousin, 2nd cousin and other cousins respectively. In this study, the most frequent type of marriage was between first cousins.

46% out of 100 families were belonged to non-consanguineous marriages that have 224 children, 134 male and 90 were female. Only 2 (0.89%) children were suffered from congenital abnormalities in non-consanguineous group.

Group.

The prevalence of Congenital Malformations in consanguineous and non-consanguineous marriage is shown in Table 2. There was an increased rate of anomalies in the consanguineous group as compared to the non-consanguineous group.

DISCUSSION

Most children who are born with major congenital anomalies and survive infancy are affected physically, mentally or socially and can be at increased risk of morbidity due to various health disorders.

In the present study, 54% of the parents belonged with consanguinity with a rate of 24%, 17% and 13% with 1st cousin, 2nd cousin and others cousin respectively. The rate of malformation was 8.2% and 0.83% in consanguineous and non-
consanguineous marriages respectively. The result of our study is in accordance with the result of the study in South East Asia which shows the prevalence of consanguineous marriages is ranging from 20-70%.

The result of our study is also in accordance with a recent “Pakistan demographic and health” survey that has shown that 2/3 of marriages in Pakistan are consanguineous marriages and nearly half of them married to the 1st cousin. Although, the rates are a bit low, as our study only reflect the situation in Khyber Pakhtunkhwa as compare to the whole Pakistan situation. Our study also shows the latest situation because with passage of time the awareness level is increases.

The prevalence of congenital anomalies at birth in developed countries is reported to be between 3-5%, those reported in Taiwan are said to be approximately 4.3%, 7.92% reported for the United Arab Emirates, 2.46% for Oman, 2.7% for Bahrain, and 3.6% for India.

However, studies in some cities of Iran such as Tehran, Yazd and Gorgan found a birth prevalence of congenital anomalies 2.3%, 3.75 and 1.01% respectively.

While in the present study, the prevalence of congenital anomalies in total numbers of children of given birth by the couples of Consanguineous Marriages is 8.2% with 3.9% in 1st cousin, 3.2% in 2nd cousin and 1.1% in others cousin marriages. This is in accordance with 7.92% reported for the United Arab Emirates. The prevalence of congenital anomalies in our study is higher as compare to most of the above studies. This is because that they reflected the prevalence of congenital anomalies only in single newborn child of the Consanguineous couple given birth in hospitalized mothers during delivery while our study reflected the prevalence of congenital anomalies in total numbers of children of the Consanguineous couple as the data was collected from the community and therefore, is more reliable.

CONCLUSION

With our study we come to the conclusion that consanguinity play an important role in the high rates of malformation seen in their offspring and must be taken into account for genetic counseling in Pakistan because collectively those abnormalities results in significant burden of diseases in the community and also on country’s economy. Furthermore, prevention is the only way of reducing the rate of congenital malformations as no effective treatment is available for majority of these cases.

RECOMMENDATIONS:

Prevention is the only way in reducing the collective burden of congenital malformations as treatment is not available for majority of these diseases at present.

Government should make compulsory checking of “Marital compatibility” before wedding of the boy and girl by law.

A subject of genetic counseling should be introduced at the school / college curriculum for awareness of the general population.

The health department may involve through doctors, nurses, paramedics, LHV’s and LHW’s of national program to enhance awareness of the people regarding the benefit of checking of “Marital compatibility” before wedding.

Media is playing an important role in educating people about the importance of “Premarital consultation / counseling” of the boy and girl. So the role of media should be insured by publishing various educating material.

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