Original Article

Association of consanguineous marriages with congenital anomalies

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Abstract:

Congenital anomalies are a major health problem all over the world; especially it is important cause of deaths and birth defects, chronic illness and disability in infants. The major cause of this is consanguineous marriages. Generation of cousin marriages have significant association with congenital anomalies **Objective:** To find out the association of consanguineous marriages with congenital anomalies present at the time of birth **Methods:** A cross sectional study was conducted at District Head Quarter Hospital, Okara from May to August, 2018. 100 adult individuals aged between 19 to 55 years, with and without cousin marriage of both genders were consecutively enrolled. Participants were assessed through pre-tested questionnaire, with prior written informed consent. Unwilling married individuals and individuals from other hospitals were not selected **Results:** According to results there was a significant association between generation of cousin marriages with congenital anomalies present at the time of birth , as p value was 0.002 **Conclusions:** Study concluded that the generation of cousin marriages has significant association with congenital anomalies present at the time of birth and due to cousin marriage 59% of the couples had congenital abnormalities in their children and 85% had genetic disorders.

Key words:

Birth defects, Congenital anomalies, consanguineous marriages

Introduction:

Consanguinity is a mixture of two words taken from Latin language; “con” means similar and “sanguineus” means blood, it indicates an association between people who are having an identical forefathers or belong to same blood [1]. Kindred ship is often referred as an association shared by two people who are biologically related to each other [2]. The rate of cousin marriage is very high in Pakistan, about 62% marriages are consanguineous. 30 million people among 200 million inhabitants of Pakistan are suffering from different forms of hereditary diseases [3]. One-fifth of population of the world, particularly the residents of North Africa, Middle East and West Asia and also the people who have migrated to other areas like North America, Australia and Europe are consistent in following the family tradition of cousin’s marriage [4]. Large numbers of cousin marriages are seen in several Arab countries, most particularly consanguineous marriages happen between first cousins, which contribute to about 25-30%. In the modern generation of few nations like Yemen, Qatar and United Arab Emirates, the cases of cousin marriages are more and are constantly enhancing [5].

Cousin marriage or kindred marriage is a major factor of progenitive problems including infant death, miscarriages, fetal death. Cousin marriage is the eminent cause of genetic disorders that are being transferred from parents to children and massive birth defects [6]. Cousin marriage is the contributing factor of congenital abnormalities. Infant mortality, fetal deaths are the consequences of consanguineous marriages. Cousin marriage may also cause

inborn heart disorders. Inborn cardiovascular malformations are widespread, affecting 2.4 to 8.0 per 1000 infants [7]. Imperfect formation of an embryo or abnormal growth may cause formational deformities in fetus referred as birth defects.

Racial, geological, cultural variations of different regions 2 are the basic factors that affect the prototype and occurrence of multiple birth defects. Different birth abnormalities occur in different countries [8]. About 1-2 % of partners become the sufferers of more than three successive pregnancy losses that happen 20 weeks prior to birth and are referred as repeated spontaneous abortions. Therefore, notionally increasing events of cousin marriages are the major factors of repeated spontaneous abortions [9]. Contamination, feed, physical condition of a mother, contagious disorders and short term ecological variations in nature together with contemporary usage of drugs are the factors of environmental changes which may also disturb gene frequency and result in congenital abnormalities [10].

Pakistan is a country with high frequency of consanguineous marriages and the congenital abnormalities are very high in off springs of such couples. The current study was aimed to highlight such abnormalities present at the time of the birth in babies born to parents who are consanguineously married. Hence, the researches will try to fill the gap in existing knowledge. The purpose of this study is to evaluate the association of genetic factors with congenital abnormalities in the population of Okara.

Methods:

A cross sectional study was conducted at DHQ Hospital, Okara from February to May, 2018. All adult individuals aged between 19 to 55 years with and without cousin marriage of both genders were consecutively selected. Signed informed consent was obtained from couples. Data were collected through non-probability convenient sampling technique and 100 participants were taken randomly. The inclusion criteria was all adult individuals, with and without cousin marriage of both genders were included and the exclusion criteria was non-cooperative individuals. Individuals were assessed through pre-tested questionnaire. Questionnaire was made according to the study objective and was pretested among 10-15 individuals, and was modified accordingly. SPSS version 21.0 was used for data analysis. Frequencies were derived and Chi-square test was applied to find out the association, p value less than 0.05 was considered significant.

Results:

Demographics of 100 participants are shown in Table 1. Analysis revealed that 28 mothers were undergraduate. Results showed that 33 women were obese as with high BMI above 30. Residential status of 77 people has their own house and 88 of the couple had marriage with their first cousin. According to results mean age was 29.85 years and mean weight was 78.9 kg (Table 1). According to results 85 of the women had familial history of any genetic disorder, 59 had congenital abnormality in children, 58 of the women faced difficulty in conceiving, and 14 had congenital abnormalities in mothers (Figure 1). According to results, 60 women were on hormonal medicines, 59 were on oral contraceptives, 76 had excessive weight gain, 58 were anemic and 65 were vitamin deficient (Figure 2).

Socio demographic characterization	Number (n)
Mothers education	
Undergraduate	28
Mothers BMI	
30.0-34.9	33
Residential status	
Own	77
Cousin marriage	
1st cousin	88
Mean Age	29.85
Mean Weight	78.89

Table 1: Socio Demographic characterization

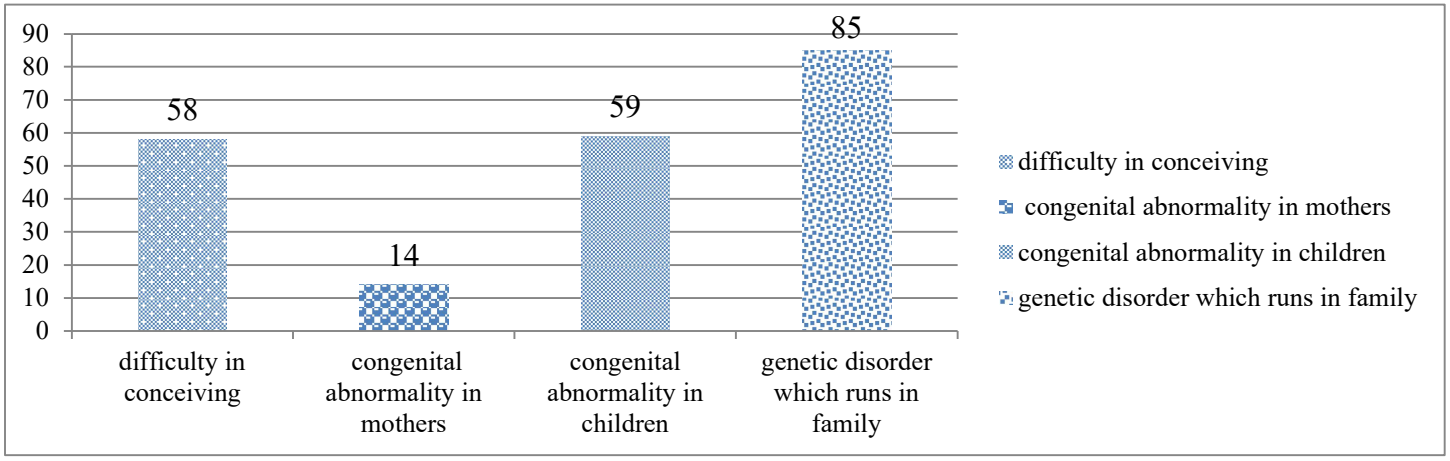

Figure 1: Congenital anomalies

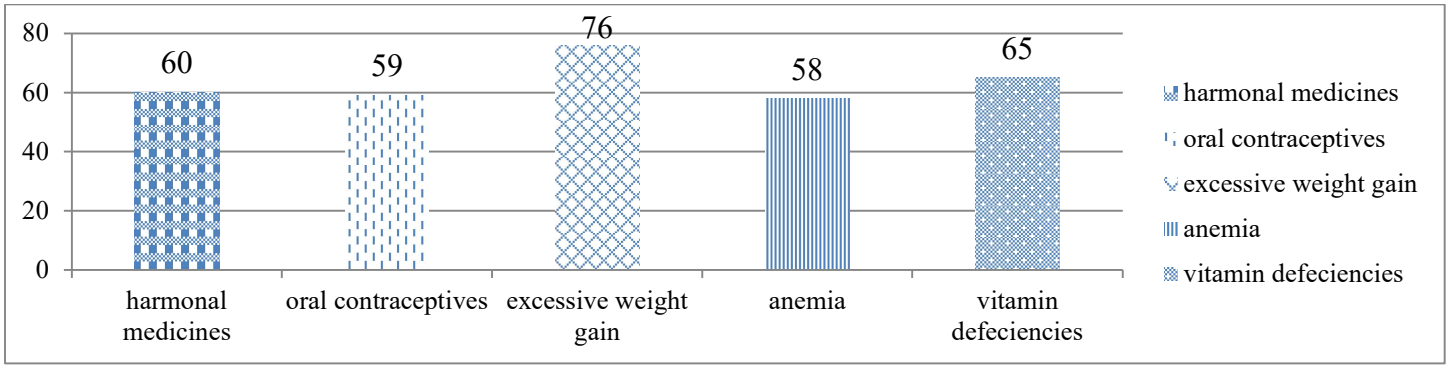

Figure 2: Risk factors and maternal health factors for congenital anomalies

Discussion:

A study was conducted to determine awareness about congenital abnormalities among pregnant women selected through non probability convenient sampling technique. According to the present study, results showed that 64% of mothers had knowledge about congenital abnormalities. Similar results were observed by a previous study conducted by Masoumeh P *et al*, (2015) to determine the risk factors and awareness of congenital anomalies in pregnant and consanguineous women. They also reported that patients who were educated had adequate knowledge about the abnormalities [11].

The results of the current study showed that 33% of mothers were obese, 27% of mothers were lying in the category of obesity grade 1, 2% were in obesity grade 2. Increase in weight is also a major factor causing problems in conceiving. Similar results were found by Kiel DW *et al*, during (2007) they were aimed to find the obesity related to mother and compared the obese and overweight pregnant females with normal weight and lean females. 59% of mothers were obese, 25% were in grade 1, and 16% were in grade 2 category. The prevalence among overweight and obese females was high than lean females [12].

According to the present study, results showed that 14% of mothers had congenital abnormalities from consanguineous marriages. Similar results were observed by Mosayebi Z *et al* during 2007 [13]. Shawky RM *et al*, during 2013 examined the effect of consanguinity on different types of genetic diseases and child morbidity and mortality. They stated that it was notably higher in many genetic diseases which suggests that couples may have deadly lethal genes, inherited from common ancestor and when transmitted to their offspring's, they can

lead to prenatal, neonatal, child morbidity or mortality [14]. Khan A *et al,* (2015), stated that congenital abnormalities are a major cause of perinatal and premature deaths [15]. The study by Singh S *et al,* in (2015) reported that functional or physical imperfections present at birth may lead to mental or physical disabilities. The result shows that central nervous system anomalies were the most prevalent congenital anomalies perceived [16].

In current study, results revealed that 88% of patients had first cousin marriage and 12% of patients had second cousin marriage. Previous study conducted Kanaan ZM *et al,* showed that the prevalence of consanguinity was 56% with 1st cousin marriage and was highest than second cousin marriages 52. Prevalence of consanguineous marriages was describing to be 42% with first cousin marriage [17].

According to the present study 26% of patients had children with abnormalities of brain, 20% of patients had children with heart problems, 13% of patients had children with cleft lip and cleft palate and 41% of patients had children with no congenital abnormality. Previous study conducted by Tomatır AG *et al.*, showed that 12 cases of brain abnormalities and 9 cases of heart disease were observed 33. Study found that anomalies of the central nervous system were the most common defect 31%, cleft palate/lip 19% and 56% were normally delivered with no congenital abnormality [18].

According to current results, 14% of patients were having congenital anomalies. In a study by Sarkar S *et al,* they determined the congenital anomalies and their risk factors in newborns. According to their results 2.22% were having congenital anomalies [19]. According to the present study, 65% of patients had miscarriages in consanguineous marriages. A previous study by Mosayebi Z *et al* during 2007 found that miscarriages reported in their study were 1.5 and 2.3 times more common in consanguineous than non/consanguineous groups respectively [13].

According to the recent study, 59% of patients were consuming oral contraceptives and 41% of patients were not consuming oral contraceptives. Similarly, Headley J *et al,* also reported that 92.4% of women used oral contraceptives at least once. The results showed that use of oral contraceptives was high during pregnancy [20].

Conclusions:

Study concluded that there was fairly increased level of congenital anomalies associated with consanguineous marriages. Majority of mothers had 1st generation cousin marriage and had difficulty in conceiving. More than half of the mothers having cousin marriage revealed that their child had anomalies such as cleft lip and plate, heart problems and abnormalities of brain. A wide range of genetic disorders were seen in families having cousin marriages. Mothers usually had taken hormonal medicines and oral contraceptives and had faced excess weight gain during pregnancy. There was a history of sudden fetal and sudden infant death. There should be increased awareness among families regarding the negative impact of cousin marriage. Health care centers also needed to educate people regarding the negative role of cousin marriages leading to abnormalities in child.

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