ORIGINAL ARTICLE

# PREVALENCE OF CONGENITAL DISEASES AMONG CONSANGUINEOUS MARRIAGES IN DISTRICT PESHAWAR PAKISTAN

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**ABSTRACT**

**Objective:** To determine prevalence of congenital diseases among the consanguineous marriages.

**Material & Methods:** A cross sectional study was carried out at the outpatient departments of the different private and public hospitals of Peshawar from November 2019 to April 2020. A total of 230 patients were interviewed using a closed ended questionnaire to obtain information on several dimensional effects of cousin marriages. The data was analyzed using SPSS 25.

**Results:** A total 230 subjects participated in the study. Parents of 169 (73.5%) children had consanguineous marriage. 11(4.8%) children had Down Syndrome followed by congenital heart defects (n=13, 5.7%), thalassemia (n=190, 82.6%), mental retardation (n=5, 1.7%) and Charcot Marie Tooth Syndrome (n=12, 5.2%).

**Conclusion:** Many patients suffered from the adverse effects of cousin marriage. It was concluded that lack of education, awareness, poverty, emphasis on cultural values were the reasons that had encouraged cousin marriages.

**Key Words:** Anomalies, Birth, Consanguinity, Education.

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## INTRODUCTION

A cousin marriage is one in which the spouses are cousins (Those who have common grandparents or another very recent ancestor).[1] According to research regarding for explanations to the higher than predicted rates of mortality and congenital abnormalities in the Pakistani community's new-borns, marriage between first cousins doubles the chance of children being born with birth problems.[2]

Children whose parents are close blood relatives are more likely to inherit genetic diseases. According to studies of cousin marriages conducted across the world, the chances of sickness and premature mortality are three to four percent greater than in the general population. Consanguinity is a well-known risk factor for genetic illnesses such as diseases and syndromes characterized by intellectual and developmental impairments. This is because to autosomal recessive diseases as well as other hereditary conditions.[2] Many diseases and syndromes can arise as a result of cousin marriage, but we were focused on the 1. Charcot Marie tooth disease (CMT) 2. Thalassemia 3. Congenital cardiac defect 4. Down syndrome 5. Mental illness. The genetic disorders of such children from consanguineous marriages resulted twice as compared to non-consanguineous marriage parents.[3,4]

In 2007, different research looked at consanguinity and the incidence of thalassemia. Each year, an estimated 5000-9000 children are affected by this disease, and the estimated carrier population percentage is 5-7%, 9% compared to a global frequency of 1%. In the province of

Punjab, where it was at 6%. The scientists said that all couples who have children should have blood testing for thalassemia and other congenital anomalies in order to reduce the chance of being intellectual and physical impairments for future.[4] The prevalence of cousin marriage throughout the world is estimated to be approximately 20%, and this rate varies from 20% to 60% in North Africa, the Middle East and Western Asia.[5] In particular, family-based close-relation CM level is 32–34% in Yemen, 29–33% in Iraq, 25–30% in Afghanistan, 14–24% in Egypt, 7–52% in South India, 44–49% in Sudan and 17–38% in Pakistan, respectively.

Study indicates that 50% of marriages in urban areas of Pakistan are between blood relatives, with 80% of these being between first cousins. Additionally, malnourished children are higher among consanguineous parents compare to non-consanguineous.[6] Earlier studies found that women married to their blood relatives experienced a greater amount of pregnancy loss, still birth and infant and childhood mortality as compared to those women married to their distant relatives or non-relatives.[7] With this background, the current study objectives were to estimate the prevalence of consanguineous marriage with congenital anomalies and also with associated factors of demographic characteristics.

## MATERIAL AND METHODS

Cross sectional study of descriptive type was carried out in District Peshawar from November 2019 to April 2020. Place of study were Hayatabad Medical Complex, Lady Reading Hospital, Fatimid Foundation, Hamza Foundation, Pak International Medical College (Pathology lab). Minimum sample size was determined by using statistical formula of Cochran's. A questionnaire contains both open ended and close ended questions. It was designed from the existing literature form "A" including demographic characteristics of the population (name, age, institution name, socio economic status, parent's education, parent's occupation, parents age, no of children, age of marriage and father and mother age at the time of marriage). The form "B" included 13 questions regarding of congenital abnormalities. The question was validated through face validity and content validity. Inclusion criteria were population suffering from the congenital birth defects due to cousin marriages. Exclusion criteria of sample Population suffering from congenital birth defects who were not married with cousin. Pilot study was not conducted because of the sensitivity of the sample. Before going to the field, institution provided the authority letter for the selected institutions. The letter was assigned to the Directors of the hospital for the pathology and other OPD department. The data was analyzed through Statistical Package for the Social Sciences (SSPS version 25). Frequency and correctional test were applied.

## RESULTS

A total of 230 participated in the study. Age categories of the participants was 0-5 years n=83(36.1%), 5-10 years n=56(24.3%),10-15years n=37(16.1%),15-20 years n=23(10.0%), 20-25 years n= 21(9.1%), 25-30 years n=10(4.3%) respectively. Majority of the subjects participated from HMC (n=88, 38.3%) followed by LRH (n=41, 17.8%), Hamza Foundation (n=75, 32.6%), PIMC (n=12, 5.2%), Fatimid Foundation (n=14, 6.1%) respectively. Analysis of socio-economic status of the participants showed that some (n=110, 47.8%) of the participants had <15000 monthly income, 74(32.2%) had 16000-50000 monthly income. Mothers of 169 (73.5%) children married to cousins. Majority n= 119(51.7%) married to 1st cousin, followed by 2nd Cousin (n=41, 17.8%), 3rd cousin (n=11, 4.8%). Most (n=95, 41.3%) of the children's father were illiterate, 42(18.3%) had elementary education followed by, Intermediate (n=23,10.0%), Bachelor (n=26, 11.3%) and University level (n=44, 19.1%). Majority (n= 144, 62.6%) of children's mother were illiterate while (n=30, 13.0%) had elementary education, 11(4.8%) had intermediate education followed by Bachelor (n=23, 10.0%), and University (n=22, 9.6%) level education. 10 (4.3%) children's' father were Doctors followed by Engineers (n=15, 6.5%), Lawyer (n=5, 2.2%), Businessman (n=50, 21.7%), and labours (n= 150, 65.2%). 11(4.8%) children had Down Syndrome followed by congenital heart defects (n=13, 5.7%), thalassemia (n=190, 82.6%), mental retardation (n=5, 1.7%) and Charcot Marie Tooth Syndrome (n=12, 5.2%). (**Table 1**).

**Table 1: Congenital anomalies of Cousin marriage and non-cousin marriage (N=230)**

| Congenital Anomalies | N | Cousin Marriage | Birth Defects/congenital anomalies | Total |
|---|---|---|---|---|
| **Down Syndrome** | 11 (4.5%) | 6 | 5 | 11 |
| **Congenital Heart defects** | 13 (5.7%) | 7 | 5 | 13 |
| **Thalassemia** | 190 (82.6%) | 142 | 48 | 190 |
| **Mental Retardation/ cleft palate** | 4 (1.7%) | 3 | 2 | 5 |
| **Charcot Marie tooth syndrome** | 12 (5.2%) | 11 | 1 | 12 |
| Total | 230 (100%) | 169 | 61 | 230 |

## DISCUSSION

A cousin marriage is one in which the spouses are relatives (i.e. people with common grandparents or people who share other fairly recent ancestors). The practice was popular in past times and continues to be common in some countries now, however such weddings are illegal in other jurisdictions. More than 10% of marriages worldwide are between first or second cousins1. The objective of current study is to determine the prevalence of diseases of cousin marriages. This study reveals that majority of patients who were suffering from defects were mostly thalassemia ages mostly between 0-5 months.169 out of 230 were married to their cousin. The frequency of people was mostly married to their 1st cousin containing maximum number of abnormalities. The illiteracy rate among mother is high. The result also showed that male are more suffers than female. Research done in Faisalabad, which included 300 consanguineous people. It represented congenital abnormality (Thalassemia) with the greatest rate (34.33%) among children aged 3 to 5 years. It was also shown that males were substantially (P<0.001) more impacted than females. While our study including 230 subjects on consanguineous, the age group affected the most was 0 - 5 years having result of 83(36%).[8] Study was conducted on new-borns at Shahid Sadoughi Hospital, Yazd ,Iran from April to December 2008 for a period of 9 months (n=1195). Current study took different places (HMC, LRH, Fatmeed Foundation, Hamza Foundation, PIMC) in the Peshawar City for data collection from 230 subjects. Consanguinity marriages are practiced by over one billion people worldwide, with a consanguinity rate of 20 - 50 percent, with the majority of them belonging to low socioeconomic position.[9] The present study was similar to the former study and demonstrated subjects who were belonging to poor families had an extremely high frequency of low socioeconomic status 110 (47.8 %). study conducted Beirut , Lebanon and Members of the National Collaborative Perinatal Neonatal Network (NCPNN) , 173 newborns were observed from Jan 2000 to Dec 2002 for a period of 3 years , Cousin marriages were responsible for congenital Heart defects ( CHD ) among the off springs ,these cousin marriages had a 1.8 times higher risk of having a CHD diagnosed at birth compared to those unrelated parents.[10] while current study conducted in the District Peshawar from 230 subjects the ratio was extremely high for the consanguinity demonstrating 73.5 %, while unrelated parents was 26.1 %. 759 Lebanese subjects the chances of congenital defects due to first cousin marriages were 34.7 (20.2 %). According to study conducted in Israel In young Bedouin 37% expressed unconditional positive attitude toward cousin marriage and 22% said that it is inevitable because of parents while in present study 74.3% advanced their children to marry his or her cousin and 25.2% didn't allowed it (30) According to study conducted in Israel in

young Bedouin 65 % sample said that cousin marriage are associated with genetic diseases while in present study 89.1% said that cousin marriage increase risk of congenital disease. Premarital screening and prenatal diagnosis was known to 97 (84.3%) and 88 (76.5%) parents while in present study 87% parents knew about pre-marital screening and genetic counselling while 13 % didn't know about it.[11] The current study demonstrated that birth defect/congenital abnormality. i.e. Down Syndrome n=11(4.8%), congenital heart defects n=13(5.7%), thalassemia n=190(82.6%), mental retardation/cleft palate n=5(1.7%), Charcot Marie tooth syndrome n=12(5.2%).

## CONCLUSION

There were strong association between consanguinity and congenital defects in our population in terms of mental retardation, thalassemia, Charcot Marie tooth syndrome, Down syndrome and congenital heart defects.

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