Readers Insight
Publisher of open access books and journals

PJMLS

Print ISSN: 2707-4471. Online ISSN: 2707-448X

Research Article	Pak-Euro Journal of Medical and Life Sciences
DOI: 10.31580/pjmls.v5i1.2470	Copyright © All rights are reserved by Corresponding Author
Vol. 5 No. 1, 2022: pp. 75-80	
www.readersinsight.net/pjmls	
Submission: February 15, 2022	**Revised:** March 18, 2022 **Accepted:** March 31, 2022

PATTERN AND PREVALENCE OF CONGENITAL MALFORMATION AND GENETIC DISORDERS AMONG OFFSPRING OF CONSANGUINEOUS PARENTS IN QUETTA

Ambreen Ijaz[1*], Maryam Muhammad Ali[1], Shubana Kausar[2], Khadija Saif[3], Masooma Mukhtar Hussain[1], Razia Muhammad Tawakal[1], Palwasha Tariq[4], Tahir Hameed[5], Gul Makai[1], Muhammad Masood Tariq Kiani[5], Nosheen Rafique[1], Aasia Karim[1], Zuhra Bibi[1]

[1]**Department of Zoology,** Sardar Bahadur Khan Women University (SBKWU), Quetta, Balochistan, Pakistan
[2]**Health Department,** Helpers Eye Hospital, Quetta, Pakistan
[3]**Gynaecology Unit 2,** Sandamen Provincial Hospital, Quetta, Pakistan
[4]**Department of Physical Therapy,** University of Arlington, Texas, USA
[5]**Center for Advanced Studies in Vaccinology and Biotechnology (CASVAB),** University of Balochistan Quetta, Pakistan
***Corresponding Author:** Ambreen Ijaz. E. mail: tariqkianiraja@hotmail.com

Abstract
This cross-sectional study was carried out to examine the pattern and prevalence of congenital malformation/genetic disorders among the offspring of consanguineous marriages in Quetta, Pakistan. A survey was conducted among 105 married couples from different areas of Quetta city. 89 out of 105 consanguineous couples were related as first cousins, followed by second cousin (n=16) and third cousin (n=6) couples. Consanguineous marriages were common among women who were illiterate (43.8%) and unemployed (87.6%). Highest number of child mortality (61.9%) and abnormalities (32.1%) were observed among the children of first cousin marriages. The most common congenital malformation/genetic disorders were mental retardation (24.13%), followed by diabetes (10.3%), deafness (6.8%) and thalassemia (6.8%). Among all the inter-related marriages, first cousin marriages are at highest risk to have babies with congenital anomalies. The aim of the current study was to examine the type, pattern and prevalence of genetic and congenital disorders among the offspring of consanguineous parents in Quetta.
Keywords: *Child mortality, Congenital anomalies, Consanguinity, Health problems*

INTRODUCTION

Cousin marriages are those which are held between couples who are related as second cousins or closer. The frequency of consanguineous marriages is different for different regions of the world. Factors like socioeconomic status and demographic features play very important role in the contribution of consanguinity being practiced in a society which includes religion, ethnicity, literacy rate, place of residence and political factors etc.

Offspring of parents who are not second cousin or distantly related has a negligible risk of inheriting two copies of recessive allele for a disorder, because recessive disorders occur when a person is homozygous for a disease meaning two copies of recessive mutation is necessary for the expression of disease (1). Congenital malformation are abnormalities which occur during pregnancy, at the time of birth or later in life of an infant (2).

In Asia, the pattern of consanguinity mostly occurs in low socioeconomic, rural areas and less educated communities of Bangladesh, Indonesia, Arab countries and in Pakistan (3). In contrast, most of the marriages in Europe, Australia and North America are not consanguineous. In these Western countries,

immigrants who migrated from developing countries with poor economy have been practicing the endogamy (4). According to a study in Pakistan the rate of consanguinity is about 60%, out of which 80% are first cousins. First cousin marriages are observed the highest in province of Balochistan and Punjab (5). Rate of neonatal death is the highest in Pakistan than most of the region of world. That is 46 deaths occur per 1,000 live births (6). In addition, due to congenital malformation each year in Pakistan, 700 children are born with genetic disorder including internal malformation, learning difficulties, neurological disorder, heart and kidney disease, thalassemia and cystic fibrosis (5).

The current study focused on examining the type, pattern and prevalence of genetic and congenital disorders among the offspring of consanguineous parents in Quetta. Several studies being conducted in Pakistan have revealed the association of consanguinity and recessive diseases; however they were mainly performed in Punjab and other provinces. In Quetta city yet there is limited research related to consanguinity and risk of recessive and congenital disorders.

METHODOLOGY

DATA COLLECTION

In this cross-sectional study, a survey was conducted over a period from January 2021 till end of March 2021. The sample comprised of 105 couples residing in Quetta city. All the couples had some degree of consanguineous relationship with their parents prior to their marriage. Procedure for respondents to fully understand the enquirers were suggested to explain the questions in Urdu, Persian and English languages. Family pedigrees and pattern of inheritance up to three/four generation were drawn to study the type of consanguineous relationship in the marriages.

QUESTIONNAIRE

A questionnaire was constructed in English language which was then explained to respondents in their local languages. It consisted of 20 multiple choice questions and 11 open-end questions. Respondents were asked regarding their marital status, socioeconomic, geographic and demographic characteristics, degree of consanguinity among couples in the family, the number of children died due to a disease and if any miscarriage or stillbirth had occurred. Educational level, awareness section and opinion on genetic counsellors were also part of the questionnaire. Coefficient of inbreeding were calculated for each individual and groups separately. Value of (F) was given to each individual based on the relationship between parents: 1/16 for first cousins, 1/64 for second cousins and 1/256 for third cousins. For groups the mean value of coefficient of inbreeding was also calculated, which is the sum of the product of each individual Coefficient by its frequency.

STATISTICAL ANALYSIS

Data entry and statistical analysis was done by the software Excel version (16.5).

RESULTS

The demographic data of respondents have been shown in Table I, according to which majority of the consanguineous marriages were observed among couples whose age at time of marriage were between 13-22 and 34-32 years for women and their husbands respectively. Number of illiterate women (43.8%) were highest while their husbands (36.1%) were having secondary level education with majority of women (87.6%) being housewives and (42.8%) of men were employed and the number of couples (60.9%) belonging to middle class status was the highest.

Out of the 105 families, 29 families had at least one individual who was affected with genetic disorder or congenital malformation as represented in Table II. 24.13% of the affected individuals were suffering from mental retardation, 10.3% were having diabetes, 6.8% were diagnosed with thalassemia and 6.8% had deafness. Other abnormalities (blindness, weak eye sight, epilepsy, delay development, cancer, cleft palate etc.) were less common with only 3.4% of all affected individuals in sample.

Table I. Consanguinity and demographic characteristics

Characteristics	Consanguineous marriages	
	Frequency	Percentage
Age at marriage (wife)		
13-22 years	69	66.6%
23-32 years	34	32.3%
33-42 years	2	0.95%
43-52 years	0	0%
Age at marriage (husband)		
13-22 years	14	13.3%
23-32 years	43	40.9%
33-42 years	37	32.3%
43-52 years	11	10.4%
Educational level (wife)		
Illiterate	46	43.8%
Primary	19	18%
Secondary	25	23.8%
Graduate	15	14.2%
Educational level (husband)		
Illiterate	22	20.9%
Primary	14	13.3%
Secondary	38	36.1%
Graduate	31	29.5%
Occupation (wife)		
House wife	92	87.6%
Working	13	12.3%
Occupation (husband)		
Employee	45	42.8%
Un-employee	29	27.6%
Own-business	31	29.5%
Financial status		
Low class	18	17.1%
Middle class	64	60.9%
High class	23	21.9%

Table III shows that 29 couples had offspring with congenital anomalies/genetic disorders with 32.1% present among children of first cousin couples and 20% among second cousin couples however, there was no affected individuals in the families with third cousin couples.

DISCUSSION

Consanguineous marriages might result with severe illnesses with offspring experiencing high risk of congenital malformation, early child death and several genetic disorders. Despite the fact that it increases the risk of abnormality, consanguineous marriages are still preferred and most commonly practiced in many countries including Pakistan(3).

CONSANGUINITY AND GEOGRAPHICAL/ETHNIC BACKGROUND

In Pakistan, a study conducted among 6611 women have shown that around 60% of marriages were consanguineous, out of which 80% of couples were related as first cousin and first cousin marriages were higher in Balochistan (53%) and Punjab (54.4%) than the other provinces (5). In this study we also found the prevalence of first cousin marriages much higher than any degree of consanguinity.

Several studies have suggested that prevalence and types of consanguineous marriages differ among different ethnicities, tribes and geographical regions. There are multiple demographic characteristics that influences the practice of consanguinity in population (7). In this study, out of 105 couples in the sample, 38 couples belonged to Hazara ethnic group, 24 couples were Baloch, while 27 and 16 couples were Pathan and Punjabi respectively.

Table II. Distribution of different types of congenital anomalies and genetic disorders

Abnormalities	No. of families		Affected individuals in each family		
	Frequency	Percentage	Frequency	Alive	Not alive
Weak eye sight	1	3.4%	27	27	0
Dicephalus	1	3.4%	1	1	0
Epilepsy	1	3.4%	2	2	0
Paralyse	1	3.4%	1	1	0
Mentally retard and leg problem, cleft palate	1	3.4%	3	3	0
Polydactyly	1	3.4%	4	3	1
Pulmonary disorder	1	3.4%	7	0	7
Multiple congenital malformation	1	3.4%	4	4	0
Blind	1	3.4%	4	4	0
Delay development	1	3.4%	3	0	3
Cancer	1	3.4%	5	0	5
SIDS	1	3.4%	8	0	8
Cleft palate	1	3.4%	3	3	0
Mentally retard	7	24.13%	36	31	5
Thalassemia	2	6.8%	3	3	0
Abnormal body parts	1	3.4%	2	1	1
Deaf	2	6.8%	5	5	0
Heart defect	1	3.4%	3	2	1
Diabetes	3	10.3%	18	17	1
Total	29	100%	139	105	32

Table III. Distribution of abnormalities in different types of consanguineous marriages

Cousin types	Normal Families		Abnormality in families		Total	Disease ratio
	Frequency	Percentage	Frequency	Percentage		
First Cousins	57	67.8%	26	32.1%	83	32.14%
Second Cousins	13	80%	3	20%	16	20%
Third Cousins	6	100%	0	0%	6	0%
Total					105	52.14%

CONSANGUINITY AND LITERACY LEVEL

The association between consanguinity and education level needs to be considered while studying the inbreeding in human population. Educational level of women has a great impact on the practise of consanguinity, several studies have shown that as the education level of women increases, the occurrence of consanguineous marriages has decreased. Present study also found that majority of women (43.8%) who were related to their husband were illiterate and around 87.6% were unemployed which was similar to the findings of several other studies (8, 9). A study done in Pakistan has revealed that the 52.2% of females who

were related to their husband as first cousins were illiterate, 48.9% had primary education, 43.1% and 32.5% had middle level education and secondary education respectively (5).

AGE AT MARRIAGE AND CONSANGUINITY

Several studies have shown that higher value of consanguinity is found among couples whose age at time of marriage was less. A survey conducted among Egyptian population showed that the value of consanguinity was significantly high in the age range of 15-25 years ($P<0.05$ and $P<0.001$) for both male and female than in older age classes (8). In this study, similar results were obtained. There was high prevalence of consanguinity in age range of 13- 22years and 23-32 years for women and men respectively.

CONSANGUINEOUS MARRIAGES AND HEALTH PROBLEMS

Many studies have inspected consanguinity and their health impact on progeny and it shows that inbreeding is associated with congenital malformation and genetic disorders. There is also a relationship between consanguinity and child morbidity and mortality, neonatal and childhood death (9, 10). Common congenital anomalies found in offspring of consanguineous couple are mental retardation, polydactyly, cleft palate, deafness, blindness and heart defects (11). First cousin couples double the risk of congenital malformation as the close relatedness of parents increases the chance of an offspring receiving both mutated genes for gene expression for a particular disorder (12). In this study, mental retardation was observed the highest (24.13%) among the families, followed by diabetes (10.3%) which was most common. Thalassemia and deafness were recorded as 6.8% among all the abnormalities found in sample. Child death has occurred the highest (13 individuals) among couples who were related as first cousins and following that was the second cousin with (8) individuals being deceased.

CONCLUSION

This research has shown that consanguineous marriages are mostly practiced among families that have low educational status and low family income due to unemployment. Babies with congenital anomalies, abortions and stillbirths are the hazardous consequences of consanguinity. Among all the inter-related marriages, first cousin marriages are at highest risk to have babies with congenital anomalies. To avoid such complications, people should be educated through both pre-marital and pre-conceptual counseling.

References:

1. Afzal M, Ali SM, Siyal HB, Hakim A. Consanguineous Marriages in Pakistan [with Comments]. The Pakistan Development Review. 1994;33(4):663-76.
2. Bittles AH, Black ML. Consanguinity, human evolution, and complex diseases. Proceedings of the National Academy of Sciences. 2010;107(suppl 1):1779-86.
3. Bittles AH. Consanguinity and its relevance to clinical genetics. Clinical genetics. 2001;60(2):89-98.
4. Chaman R, Taramsari MG, Khosravi A, Amiri M, Naieni KH, Yunesian M. Consanguinity and neonatal death: a nested case-control study. Journal of family & reproductive health. 2014;8(4):189.
5. Ng D, Ahern K. The implications of parental consanguinity on the Care of Neonates. Advances in Neonatal Care. 2016;16(4):273-82.
6. Hamamy H. Consanguineous marriages. Journal of community genetics. 2012 Jul;3(3):185-92.
7. Akbayram S, Sari N, Akgün C, Dogan M, Tuncer O, Caksen H, Öner AF. The frequency of consanguineous marriage in eastern Turkey. Genetic Counseling. 2009;20(3):207.
8. Veleanu C. La passion et le juridique: le crime passionnel en anglais et dans quelques langues romanes. Interstudia (Revista Centrului Interdisciplinar de Studiu al Formelor Discursive Contemporane Interstud). 2012(12):113-21.
9. Bener A, Mohammad RR. Global distribution of consanguinity and their impact on complex diseases: Genetic disorders from an endogamous population. Egyptian Journal of Medical Human Genetics. 2017;18(4):315-20.

10. Madondo A, Ntuli B, Macintyre UE. The clinical and anthropometric profile of undernourished children aged under 5 admitted to Nyangabgwe Referral Hospital in Botswana. South African Journal of Child Health. 2012;6(4):123-7.
11. Hussain R. Consanguinity: Cultural, religious and social aspects. InGenetic Disorders of the Indian Subcontinent . Dordrecht. 2004:125-135.
12. Joseph N, Pavan KK, Ganapathi K, Apoorva P, Sharma P, Jhamb JA. Health awareness and consequences of consanguineous marriages: A community-based study. Journal of primary care & community health. 2015;6(2):121-7.
13. Reza C, Mahshid Gholami T, Ahmad K, Mohammad A, Kourosh Holakouie N, Masoud Y. Consanguinity and neonatal death: a nested case-control study. 2014:189-193.