# Open Access Full Length Article

ORIGINAL ARTICLE

# Knowledge and Awareness about Genetic Problems Associated with Consanguineous Marriages among Non-Medical Students of Islamabad

# Memoona Rasheed<sup>1</sup>, Naghmi Asif<sup>2</sup>, Khalid Hassan<sup>3</sup>

<sup>1</sup> Research Officer, Department of Pathology, Islamabad Medical and Dental College. <sup>2</sup> Professor, Department of Pathology, Islamabad Medical and Dental College <sup>3</sup> Professor, Head Department of Pathology, Islamabad Medical & Dental College (Shaheed Zulfigar Ali Butto Medical University, Islamabad)

#### **ABSTRACT**

Objective: To access the level of knowledge of non-medical students about genetic problems related to consanguinity. Methodology: A descriptive study was conducted from September 2015 to June 2016 to collect information regarding acquaintance and perceptions about, consanguineous marriages among students of different colleges and universities of Federal Capital. Specially designed performa was used to collect data. The collected data was analyzed by using Statistical Package for Social Sciences (SPSS) software version 16.0. Demographic characteristics of participants were calculated as Mean±SD. Chi-square test was applied to check the level of significance among different categorical variables.

Results: A total of 424 students were surveyed, out of which 155 (36.6%) were male and 269 (63.4%) were female. The mean age of the study population was 20.39 ±2.44. The data analysis showed only 9.7% of students are fully aware of the problems associated with consanguinity. Girls were found to be more aware as compared to boys (p= 0.05) and among all ethnic groups, Punjabis were found to be more aware (16.9%). In subjects with a history of parental consanguinity and consanguinity in siblings, low level of awareness (8.8% and 8.5% respectively) was found.

Conclusion: Efforts should be made to increase the knowledge about consanguinity and its association with genetic diseases among non-medical students.

**Key Words:** Awareness, Consanguinity, Genetic diseases.

Author's Contribution	Address of Correspondence	Article info.
<sup>1</sup> Conception, synthesis, planning of research and	Memoona Rasheed	Received: June 4, 2017
manuscript writing, <sup>2</sup> Data Analysis, <sup>3</sup> Conception,	Email: memoona_rasheed@hotmail.com	Accepted: August 29, 2017
Review the Study, <sup>3</sup>	_	-

Cite this article. Rasheed M, Asif N, Hassan K. Knowledge and Awareness about Genetic Funding Source: Nil Problems Associated with Consanguineous Marriages among Non-Medical Students of Conflict of Interest: Nil Islamabad. JIMDC. 2017: 6(3): 189-193.

#### Introduction

Consanguinity, a union between close biological kin, is most common in the Middle East, South and West Asia and North Africa<sup>1</sup>. These unions vary widely from the patrilateral parallel consanguineous union in the Middle East to uncle-niece marriages in parts of South India.<sup>2</sup> Rate of intermarriages varies greatly among a population and is influenced by factors such as religion, education,

socio-economic status, geography and local tradition.<sup>3,4</sup> Due to such unions, gene flow between communities has been highly restricted resulting in a very different inherited disease profile, reflecting local founder effect and gene drift.5 The level of homozygosity is 12.5% among first cousin union while the homozygosity level in second cousin marriage is also higher from that in general

population.<sup>6</sup> Currently, 10.4% of world's population is married to a second cousin or closer (Coefficient of inbreeding  $\geq$ 0.0156).<sup>7</sup>

The rate of consanguineous marriages in Pakistani population is ~62.7% of which ~80.4% unions are between first cousins.8 The common reasons being the protection of property, close family ties, familial compatibility, low divorce rate and a part of faith.9 Prevalence of congenital recessively inherited disorders is high among consanguineous parents. 10 During the last few decades, many rare disease genes have been identified and their chromosomal location has been mapped by studying multiplex consanguineous families. These studies have led to the identification of novel disease-causing gene mutations in Pakistani families showing various abnormal phenotypes such as intellectual disability. 11 eye abnormalities, 12-14, Deafness 15 and Thalassemia<sup>16</sup>. This study is aimed to access knowledge of adolescent of Islamabad about consanguinity and its probable outcomes.

#### **Patients and Methods**

A descriptive study was conducted in Capital Territory of Pakistan from September 2015 to June 2016. A structured questionnaire was designed to collect demographic information such as age, gender, ethnicity, residence, native language, and education. Questions were included to evaluate overall knowledge of respondents about consanguinity and prevalence of consanguinity in their families. The study was approved by the Institutional Review Board and informed consent was obtained from all the participants. Students of Higher Secondary School Certification and University level were included in the study. Students of Medical colleges and of secondary school certificate were excluded from the study. The data was collected from students of various departments studying at Quaid-i-Azam University, Air University, Federal Urdu University, International Islamic University, Comsats Institute of Information Technology and various Public-sector colleges of Islamabad. The data were recorded and analyzed by using Statistical Package for Social Sciences version 16.

The questionnaire consisted of 16 questions to assess the level of knowledge of the respondents about consanguinity. Awareness was calculated by using the method described by Jaber et al,<sup>17</sup> with slight modification in numbering. Each question contributed 1 point to the final score. The students who answered 0-8 questions correctly were considered to have a low level of awareness, students who answered 9-12 questions correctly were considered to have a moderate level of awareness and students who responded 13-16 correctly, were considered to have a high level of awareness. Questions were also included to access the acceptability of cousin marriages in families of the respondents and presence of congenital disorders.

#### Results

A total of 424 students participated in this study. The mean age of the participants was  $20.39 \pm 2.44$ . More than 99% of the participants were Muslims belonging to different ethnic backgrounds.

The data analysis showed that 54.5% students have a low level of awareness, 35.8% have a moderate level of awareness while 9.7% of the students have a high level of awareness about the problems associated with consanguinity. Among students surveyed, 66% knew that consanguinity was associated with high incidence of congenital malformations in children. Among these, only 69.6% knew which congenital malformations were associated with consanguinity. (Figure 1)



Figure 1: Awareness level of the respondents about problems related to consanguinity.

Table 1 represents the demographic characteristics of the participants. Among total participants, 116 (27.4%) were enrolled students of business administration, 163 (38.4%) were from natural sciences, 67 (15.8%) were from applied sciences and 78 (18.4%) of respondents were from higher secondary school education (Table 1).

Table 1: Demographic characteristics of the participants			
Demographic Characteristic	Number		
		(Percentage)	
Gender			
Male		155 (36.6)	
Female		269(63.4)	
Religion			
Islam		421 (99.3)	
Hindu/Sikh		2 (0.5)	
Christian		1 (0.2)	
Ethnicity			
Urdu Speaking		240 (56.6)	
Punjabi		108 (25.4)	
Pashto		44 (10.3)	
Sindhi		6 (1.4)	
Balochi		5 (1.2)	
Others		21 (4.9)	
Residential Area			
Urban		356 (84)	
Sub-Urban		19 (4.5%)	
Rural		49 (11.5)	
Education			
Business Administration	BBA	80 (18.9)	
Dusiness Auministration	MBA	36 (8.5)	
	Pharm D	42 (9.9)	
Natural Sciences	BSc	79 (18.6)	
	MSc	42 (9.9)	
And all of Original	BSCS	27 (6.5)	
Applied Sciences	BSSE	40 (9.4)	
Others		78 (18.3)	
Institution			
Quaid-i-Azam University		128 (30.2)	
Federal Urdu University of Science and		93 (21.9)	
Technology			
International Islamic Universit	52 (12.3)		
Comsats Institute of Information Technology		54 (12.7)	
Air University		33 (7.8)	
Others		64 (15.1)	

Table 2 represents demographic characteristics showing an impact on an awareness level. According to the type of degree, education was divided into three groups i.e. intermediate level, undergraduate level and postgraduate level. The data analysis showed that postgraduate students have more awareness (19.8%) as compared to those enrolled in the undergraduate or intermediate programme (3.4% and 12.7%).

Table 2: Awareness level of the participants about problems related to consanguinity					
Variables	Low level of	Moderate level of	High level of	p Value	
	awareness	awareness	awareness		
Gender					
Male	104 (67.1)	46 (29.7)	5 (3.2)		
Female	126 (46.8)	107 (39.8)	36 (13.4)	0.00	
Religion					
Islam	227 (54)	153 (36.3)	41 (9.7)	0.00	
Hindu/Sikh	2 (100)	0	0		
Christian	1 (100)	0	0		
Residential Area					
Urban	196 (55.1)	126 (35.4)	34 (9.5)		
Suburban	8 (42.1)	8 (42.1)	3 (15.8)	0.00	
Rural	25 (51)	20 (40.8)	4 (8.2)		
Ethnicity	, ,	. ,	. ,		
Urdu Speaking	137 (57.1)	85 (35.4)	18 (7.5)		
Punjabi	43 (51.8)	26 (31.3)	14 (16.9)		
Pathan	14 (31.8)	29 (65.9)	1 (2.3)	0.00	
Sindhi	4 (66.6)	1 (16.7)	1 (16.7)		
Balochi	5 (100)	0	0		
Others	27 (58.7)	12 (26.1)	7 (15.2)		
Education	, ,	,	, ,		
Intermediate	38 (60.3)	17 (27)	8 (12.7)		
Undergraduate	122 (51.9)	105 (44.7)	8 (3.4)	0.00	
Postgraduate	70 (55.6)	31 (24.6)	25 (19.8)		
Parental Consan	· '				
Consanguineous	111 (54.4)	75 (36.8)	18 (8.8)	0.00	
Non-	93 (46.8)	74 (39)	23 (12.1)		
Consanguineous	, ,	, ,	, ,		
Not Known	26 (86.7)	4 (13.3)	0 (0)		
Consanguinity in					
Consanguineous	60 (56.6)	37 (34.9)	9 (8.5)		
Non-	170 (53.8)	115 (36.4)	31 (9.8)	0.00	
Consanguineous					
Congenital disorders in family					
Yes	35 (64.8)	19 (35.2)	0		
No	194 (52.6)	134 (36.3)	41 (11.1)	0.00	

Out of the surveyed students, 155 (36.6%) were male and 269 (63.4%) were female. Among the two genders, the percentage of girls with a high level of awareness was found to be significantly greater than that of boys (p=0.00). We also compared the level of education with gender. A number of female enrolled in postgraduate degree was high (31.2%) as compared to boys (27.1%) (Table 2).

However, number of boys enrolled in the undergraduate programme (72.3%) was more as compared to girls (45.7%) (Table 3).

Among all ethnic groups, Punjabi (16.9%) and Sindhi (16.7%) were found to have more awareness (Table 2). Comparison between ethnic groups and education level showed that number of students belonging to pathan community (36.4%) are higher in postgraduate level than other ethnic groups while the number of Urdu speaking students are enrolled more in undergraduate level (58.3%) (Table 3).

Table 3: Comparison between gender and ethnicity with education level					
Variable	Education Level			p- Value	
	Inter mediate	Under graduate	Post graduate		
Gender					
Male	1 (0.6)	112 (72.3)	42 (27.1)	0.00	
Female	62 (23.1)	123 (45.7)	84 (31.2)		
Ethnicity					
Urdu Speaking	36 (15)	140 (58.3)	64 (26.7)		
Punjabi	20 (18.5)	57 (52.8)	31 (28.7)	0.00	
Pathan	4 (9.1)	24 (54.5)	16 (36.4)		
Parental Consanguinity					
Consanguineous	28 (13.7)	117 (57.4)	59 (28.9)		
Non- Consanguineous	31 (16.2)	102 (53.4)	58 (30.4)	0.00	
Not Known	4 (13.8)	16 (55.2)	9 (31)		

Comparison of knowledge between students of consanguineous and non-consanguineous parents showed that even children of consanguineous parents had low awareness. (Table 2).

Students whose one or more siblings were married within the family have relatively less awareness (8.5%) as compared to those whose siblings were not married within family or cast (9.8%). The students were questioned about the presence of any congenital disorder in their siblings such as deafness, mental retardation or blindness. Of the 424 respondents, 54 (12.7%) reported the presence of at least one disorder in their siblings. Students who had siblings with congenital abnormality showed a significantly lower level of knowledge as compared to those without such abnormalities (p=0.00) (Table 2).

#### Discussion

The study results showed that almost half (54.2%) of the students were unaware of the problems associated with consanguinity. Among the two genders, girls were found to be more aware (13.4%) as compared to boys (3.2%). The probable reason for this finding can be that more female were enrolled in post-graduation level as compared to male. This finding is in line with the findings of similar studies conducted on Saudi adults, Egyptian and Israeli Arab Population. 17-19 Students belonging to urban and rural areas showed a lower level of awareness as compared to those belonging to suburban areas. Among all ethnic groups, Punjabis showed more awareness as compared to others. A number of Sindhi and Balochi respondents were less in our target population. Extending similar studies in these populations can give more details. Studies conducted in Azad Kashmir showed an increase in consanguinity in literate subjects.20

Consanguinity among parents and sibship was found to be associated with lower level of awareness. Same trends were observed in the comparison between awareness level and presence of congenital malformation. Our findings are in line with the studies conducted on Israeli, Arab adolescent.<sup>17</sup> The comparison between parental consanguinity and education level of participants showed that percentage of children belonging to nonconsanguineous parents enrolled in the postgraduate level were more (30.4%) as compared to consanguineous parents (28.9%). Ghafoor et al., 21 showed that even level of awareness about thalassemia among parents of diseases patients was low. As parents are the primary source of information for children lack of awareness of parents can be a probable reason for unawareness in their children despite the presence of consanguinity and congenital disorders.

#### Conclusion

Our finding showed an overall low of awareness about problems associated with consanguinity among non-medical students. We suggest that this topic should be considered while planning educational curriculum in secondary and higher secondary education system so that they can educate their family members and thus can

contribute to reducing the burden of such genetic diseases from our society. Moreover, awareness talks in colleges and universities and through electronic media will also reduce the percentage of consanguinity and rate of genetic diseases seen with consanguineous marriages.

### References

- Bittles AH. Consanguinity, genetic drift, and genetic diseases in populations with reduced numbers of founders. In: Speicher MR, Antonarakis SE, Motulsky AG, editors. Vogel and Motulsky's Human Genetics. Springer-Verlag.: Berlin Heidelberg; 2010. 507–528.
- Bittles AH The role and significance of consanguinity as a demographic variable. Pop Dev Rev. 1994; 20:561–584.
- Fuster V, Colantonio SE. Socioeconomic, demographic, and geographic variables affecting the diverse degrees of consanguineous marriages in Spain. Hum Biol. 2004;76:1-14.
- Kerkeni E, Monastiri K, Saket B, Rudan D, Zgaga L, Ben Cheikh H. Association among education level, occupation status, and consanguinity in Tunisia and Croatia. Croat Med J. 2006;47:656-61.
- Bittles AH, Black ML. Consanguinity, human evolution, and complex diseases. Proceedings of the National Academy of Sciences. 2010;107(suppl 1):1779-86.
- Miguel A, Alfonso-sa'nchez, Jose AP. Effects of Consanguinity on Pre-reproductive Mortality: DoesDemographic Transition Matter? American Journal of Human Biology. 2005; 17(6): 773-786.
- Chisholm JS and Bittles AH. Consanguinity and the Developmental Origins of Health and Disease. Journal of Evolutionary Medicine. 2015;3(4); Article ID 235909. doi:10.4303/jem/235909
- Hussain R, Bittles AH. The prevalence and demographic characteristics of consanguineous marriages in Pakistan. J Biosoc Sci. 1998;30(2):261–75.
- Khan HGA, Sultana A, Awais-e-Siraj. Consanguinal marriages in Pakistan: a tradition or a cultural politics and possible management measures? Canad Soc Sci. 2011;7(1):119-23.
- Ahmed S, Saleem M, Sultana N, et al. Prenatal diagnosis of bthalassaemia in Pakistan: experience in a Muslim country. Prenat Diagn. 2000; 20:378-83.

- Harripaul R, Vasli N, Mikhailoy A, Rafiq MA, et al., Mapping autosomal recessive intellectual disability:combined microarray and exome sequencing identifies 26 novel candidate genes in 196 consanguineous families. DOI: https://doi.org/10.1101/092346.
- Ravesh Z, El Asrag ME, Weisschuh N, et al. Novel C8orf37 mutations cause retinitis pigmentosa in consanguineous families of Pakistani origin. Molecular Vision. 2015;21:236-243.
- Rauf B, Irum B, Kabir F, et al. A spectrum of CYP1B1 mutations associated with primary congenital glaucoma in families of Pakistani descent. Human Genome Variation. 2016;3:16021-. doi:10.1038/hgv.2016.21.
- Ullah E, Saqib MAN, Sajid S, Shah N, Zubair M, Khan MA, Ahmad I, et al., Slayotinek A. Genetic analysis of consanguineous families presenting with congenital ocular defects. Experimental Eye Research, 2016, 146: 163-171.
- Rehman AU, Santos-Cortez RLP, Morell RJ. Mutations in TBC1D24, a Gene Associated With Epilepsy, Also Cause Nonsyndromic Deafness DFNB86. AJHG. 2014; 94(1): 144– 152.
- Yasmeen H, Toma S, Killen N, Hasnain S, Foroni L. The molecular characterization of beta globin gene in thalassemia patients reveals rare and a novel mutations in Pakistani population. 2016;59(8):355-62.
- 17. Jaber L, Shohat M, Halpern GJ. Demographic characteristics of the Israeli Arab community in concentration with consanguinity. Isr J Med Sci 1996; 32(12): 1286-1289.
- Ahmed AE, Alharbi OA, Al-Hamam AA, Al-Shaia WA, Al-Marzoug HM, Bagha M. Awareness of Health Consequences of Consanguineous Marriages among Saudi Adults. Jphdc. 2016, 2(1):121-129.
- Shelkamy FR, Alaaef-Din S, Kotb SA, Sharkawy SA. Knowledge and attitude of students living in assiut university dorms about consanguinity. The medical journal of Cairo University, 2013;81(2):39-46.
- 20. Jabeen N, Malik S. Consanguinity and its socio-demographic differentials in District Bhimber, Azad Jammu Kashmir, Pakistan. J Health Pop Nutr. 2014;32(2):301–13.
- Ghafoor MB , Leghari MS , Mustafa G , Naveed S. Level of Awareness about Thalassemia among Parents of Thalassaemic Children. JRMC. 2016; 20(3):209-211.