

## **Prevalence and Associated Factors of Genetic Disorders and Congenital Abnormalities in Consanguineous Marriages in Afghanistan**

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### **Abstract**

**Background:** Consanguineous marriage, the union between two individuals closely related, is a prevalent practice in Afghanistan, with an estimated 46.2% of all marriages being consanguineous. This practice is driven by various factors, including cultural traditions, beliefs in strengthening familial bonds, and ease of finding a suitable spouse within the extended family. However, consanguineous marriages have been associated with an increased risk of genetic disorders in offspring. This study aimed to investigate the prevalence and associated factors of genetic disorders and congenital abnormalities in a group of couples with consanguineous marriage in Afghanistan.

**Methodology:** A cross-sectional study design was employed to recruit 131 couples with consanguineous marriage from various hospitals in Kabul, Afghanistan. Participants were selected on questionnaire from registered patients admitted to the hospitals based on their marital status, consanguinity, and reasons for visiting the doctor, including marital problems, childlessness, repeated abortions, congenital and genetic diseases in their children. Data were collected on demographic characteristics, medical history, physical examination findings, and genetic testing results and descriptive statistics (mean, median, mode, standard deviation, variance, frequency) conducted.

**Results:** The study findings revealed that 30% of the participating couples had children with genetic disorders. Among these couples,

59% of the cases were associated with first cousin marriages. The most common type of consanguineous marriage was first cousin marriage, accounting for 61.8% of all consanguineous marriages. These findings highlight the significant association between consanguineous marriage and the prevalence of genetic disorders in Afghanistan.

**Conclusion:**

This study delves into Afghanistan's common practice of consanguineous marriage, particularly first cousin unions (over 60%). Worryingly, it reveals a stark link between this practice and an increased risk of children with genetic disorders. While details require further exploration, these findings highlight the crucial need for accessible genetic counseling and education to empower couples to make informed choices regarding their reproductive health and future generations.

**Keywords:** Consanguineous Marriage, Genetic disease, Afghanistan, Cultural Practice

## Introduction

Consanguineous marriage, also known as cousin marriage, is a union between two people who are closely related like have a common ancestor or blood, typically second cousins or closer. It is a common practice in many parts of the world, particularly in the Middle East, South Asia, and North Africa [1, 2]. Approximately 1.1 billion people with consanguineous marriages currently live in world countries where family marriage is common [1].

In Afghanistan, the number of consanguineous marriage is 46.2% of all marriages [3], with the highest number of marriages in Bamyan with 51.2%, followed by Kabul with 38.2%, compared to neighboring countries such as Pakistan, which has almost 65% consanguineous marriages in all its marriages, followed by India (55%), Saudi Arabia (50%), Iran (30%), Egypt, and Turkey (20%) [4, 5].

The inbreeding coefficients in Afghanistan are some of the highest in the world. The most common type of cousin marriage in Afghanistan is first cousin marriage, which accounts for 27.8% of all marriages. This is followed by marriage to two first cousins (6.9%), second cousin marriage (5.8%), and marriage to a first cousin once removed (1.8%). First cousins can be children of two brothers (parallel paternal cousins), children of two sisters (material parallel cousins), or children of a brother and sister (cross cousins) [5, 6].

Family marriages, or marriages between close relatives, are driven by various factors. One prominent reason is the belief that such unions strengthen familial bonds and unity, fostering a sense of togetherness and mutual support during challenging times. Additionally, deep emotional connections between siblings may lead to their children marrying each other [7-9].

The ease of finding a suitable spouse among family members is another determining factor. Parents often find it simpler to assess the financial stability, career prospects, and compatibility of potential partners within their own extended family [10, 11]. The close proximity of families, particularly in rural areas, also plays a role. Families often prefer their children to remain close after marriage, leading to a preference for consanguineous unions [10, 12].

Furthermore, divorce rates in consanguineous marriages are generally lower. In the event of marital disputes, both families can often intervene and resolve issues amicably. Additionally, families

may exert pressure on the couple to work through their differences and remain together [7, 10].

Dowry and inheritance also play a significant role in prompting family marriages. Keeping property and land within the family is a primary concern, and consanguineous unions prevent the division of assets. Some families also believe that a daughter-in-law from within the family will provide support in their later years [7, 13].

Recent studies have revealed a heightened susceptibility to certain diseases among children born from consanguineous marriages. For instance, one study found a thalassemia incidence of 5% in the general population compared to 32% in offspring from consanguineous unions [14, 15]. A study by researchers from National Association of Genetic Counselors found that children of first cousin marriages in North America, Africa, Asia, and the Middle East have a 7-8% risk of developing recessive genetic disorders such as Tay-Sachs disease and cystic fibrosis [14, 15]. Other diseases linked to consanguineous marriages include autosomal recessive genetic disorders like cystic fibrosis and sickle cell anemia, congenital anomalies such as hydrocephalus, polydactyly, and cleft lip and palate, mental health disorders like bipolar disorder and depression, fertility issues including infertility, increased infant and child mortality, congenital heart defects, and Down syndrome [15]. This study aimed to investigate the prevalence and associated factors of genetic disorders and congenital abnormalities in a group of couples with consanguineous marriage in Afghanistan.

## **Methodology**

### **Study Design**

This study employed a community-based cross-sectional design to investigate the prevalence and associated factors to investigating the prevalence of genetic diseases and congenital

abnormalities in a group of couples with consanguineous marriage in Afghanistan

### **Study Population**

A total group of 131 couples with consanguineous marriage aged 22-54 years out of 250 couples were included in this study. Participants were selected from registered patients admitted to various hospitals in Kabul, who met the inclusion criteria, the reason for

visiting the doctor was due to marital problems; childlessness, repeated abortions, congenital and genetic diseases in their children, and consanguineous marriage by healthcare professionals.

### **Data Collection**

**Sampling:** The data was collected based on questioner. A systematic random sampling technique was employed to select participants from the list of registered patients in selected hospitals. A sampling interval was determined, and every patient on the list was included in the study.

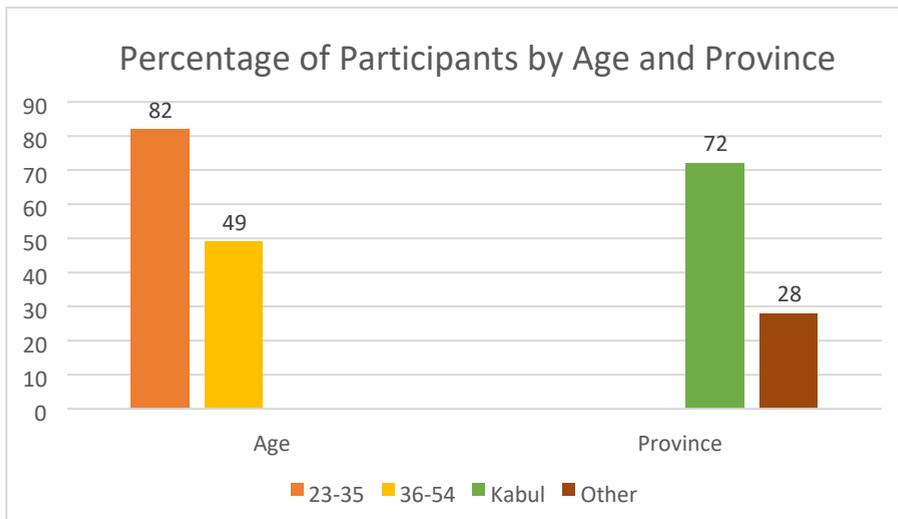
- Collect demographic data on all participants, including age, sex, ethnicity, education level, and occupation.
- Collect detailed medical history on all participants, including past medical history, family medical history, and current medications.
- Perform physical examinations on all participants to assess for any signs of genetic disorders or congenital abnormalities.
- Collect genetic samples from all participants for genetic testing.
- Perform genetic testing for a panel of known genetic disorders associated with consanguineous marriage.

### **Data Analysis**

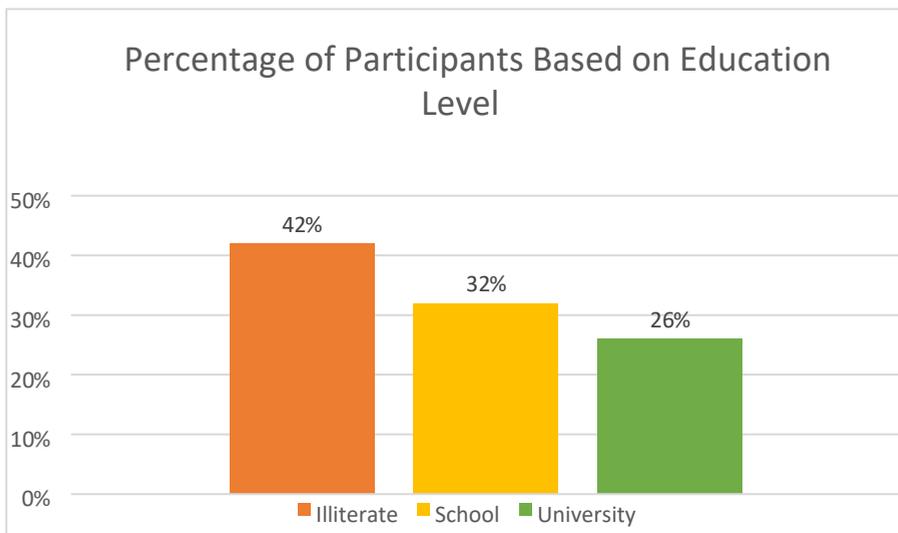
- The demographic and clinical data were entered into SPSS Statistics 26 and descriptive statistics (mean, median, mode, standard deviation, variance, frequency) conducted to identify any associations between consanguineous marriage and the prevalence of genetic disorders and congenital abnormalities.
- Analyze genetic data to identify any genetic variants associated with the observed phenotypes.

### **Result**

The research shows that from 131 couple participated in this study, the majority of participants (62%) are in the 23-35 age group, followed by the 36-54 age group (37%). An Also (54%) of participants are from Kabul province, followed by Balkh, Ghazni, Bamyān, Kandahar, Daikundi, Logar and Herat provinces (46%).

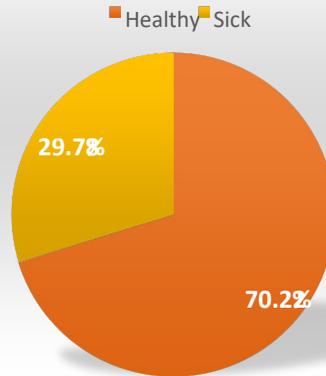


The majority of the study participants ( $n=55$ ) have no formal education. This is followed by those with a secondary education ( $n=42$ ), and the smallest group is those with university education ( $n=34$ ).



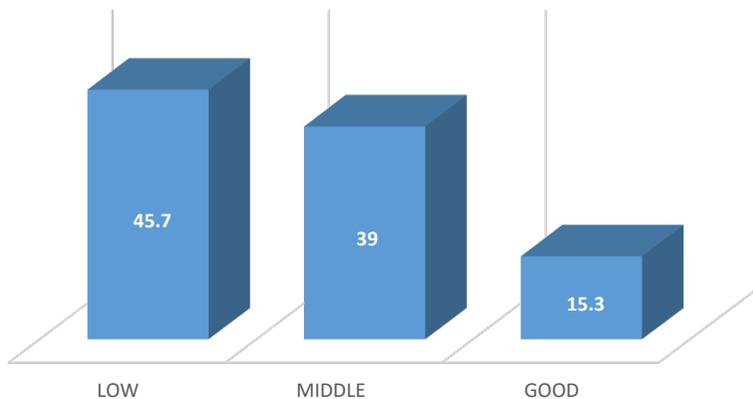
Out of the 131 couples participating in the study, 39 couples had children with genetic disorders, while the remaining 92 couples had unaffected children

### Percentage of Participants based on having Children with Genetic Disease

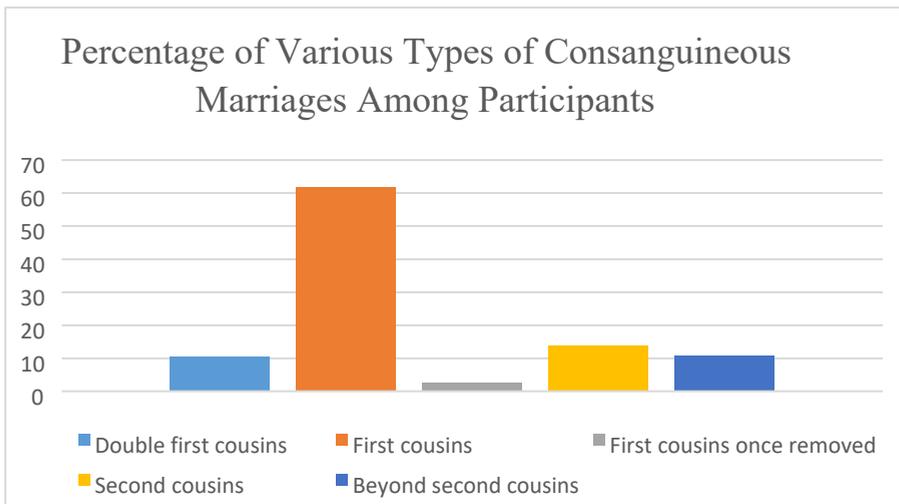


Among 39 couples having children with genetic disorder; 18 were with lowest income household, followed by 15 with middle and 6 with good incomes.

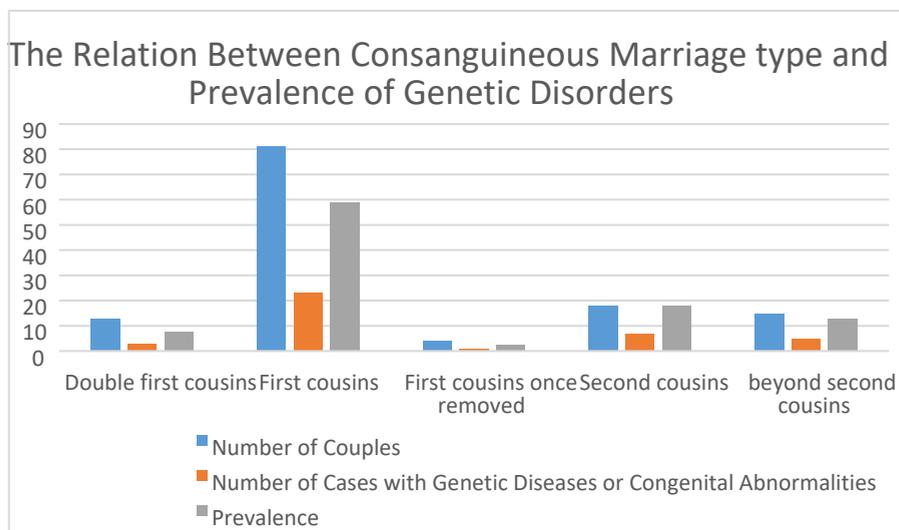
### PERCENTAGE OF INCOMES RELATED TO GENITIC DISORDERS



The most common type of consanguineous marriage among participants in this study is first cousin marriage 81 marriages (61.8%). This is followed by second cousin marriage 18 marriages (13.77%), beyond second cousin marriage 15 marriages (10.71%), double first cousin marriage 13 marriages (10.4%), and first cousin once removed marriage 4 marriages (2.6%).



In a study of 131 couples, 39 of couples had children with genetic disorders. Among these couples, 23 (59%) of the cases were associated with first cousin marriages, 7 (18%) with second cousin marriages, 5 (12.9%) with marriages beyond second cousin relationships, 3 (7.7%) with double first cousin marriages, and 1 (2.4%) with marriages between first cousins once removed.



## Discussion

This study reveals a concerning link between consanguinity and genetic disorders. Over 60% married first cousins, with higher rates of affected children linked to closer genetic relations. Notably, lower socioeconomic status correlated with increased risk. Education, genetic counseling, and accessible prenatal screening empower families and improve health outcomes.

The study provides valuable insights into the prevalence of consanguineous marriage in Afghanistan and its association with genetic disorders. The findings reveal that consanguineous marriages are common, particularly first cousin marriages, and they carry a significantly increased risk of genetic disorders in offspring [16, 17]. This is consistent with previous studies from other regions with high rates of consanguineous marriage.

The study's findings have important implications for public health and genetic counseling in Afghanistan. Consanguineous marriages are deeply ingrained in Afghan culture and traditions, and they often hold social and economic benefits. However, the increased risk of genetic disorders associated with consanguineous marriages cannot be ignored [13, 16].

Genetic counseling can play a crucial role in educating couples about the risks and benefits of consanguineous marriages, enabling them to make informed decisions about their reproductive health. Genetic counselors can provide information about the specific genetic disorders associated with different types of consanguineous marriages and the options available for prenatal and carrier screening [18, 19].

In addition to genetic counseling, public health education campaigns can raise awareness about the risks of genetic disorders associated with consanguineous marriages and promote informed decision-making among couples. These campaigns should be culturally sensitive and tailored to the specific needs and beliefs of the Afghan population [18-20].

Future research should focus on further understanding the complex factors influencing consanguineous marriage practices in Afghanistan and their implications for genetic disorders. Additionally, research should explore the effectiveness of genetic counseling and public health education interventions in addressing this issue.

### **Strengths:**

Consanguineous marriages, particularly first cousin marriages, are prevalent in Afghanistan.

Consanguineous marriages are associated with an increased risk of genetic disorders in offspring.

Genetic counseling and public health education can play important roles in addressing this issue.

Further research is needed to fully understand the complexities of consanguineous marriage practices and their implications in Afghanistan.

### **Conclusion**

This study examined the prevalence of consanguineous marriage and its association with genetic disorders in Afghanistan. The findings reveal that consanguineous marriages are common in Afghanistan, particularly first cousin marriages, which account for over 60% of cases. Additionally, the study found that consanguineous marriages are significantly associated with an increased risk of children with genetic disorders. Among couples with children with genetic disorders, the majority were associated with first cousin marriages (59%). While further research is needed to fully understand the complex factors influencing consanguineous marriage practices and their implications in Afghanistan, these findings underscore the importance of genetic counseling and education for couples considering consanguineous marriages to make informed decisions about their reproductive health.

### **Conflicts of Interest:**

The authors declare that there are no competing interests.

### **Founding**

Not founded

## References

1. Ben-Noun, L., *CONSANGUINEOUS MARRIAGES FROM ANTIQUITY TO THE PRESENT*. 2017.
2. Hamamy, H., *Consanguineous marriages : Preconception consultation in primary health care settings*. J Community Genet, 2012. **3**(3): p. 185-92.
3. Saify, K. and M. Saadat, *Consanguineous marriages in Afghanistan*. J Biosoc Sci, 2012. **44**(1): p. 73-81.
4. Iqbal, S., et al., *Consanguineous marriages and their association with women's reproductive health and fertility behavior in Pakistan: secondary data analysis from Demographic and Health Surveys, 1990-2018*. BMC Womens Health, 2022. **22**(1): p. 118.
5. Ullah, M.A., A.M. Husseni, and S.U. Mahmood, *Consanguineous marriages and their detrimental outcomes in Pakistan: an urgent need for appropriate measures*. International Journal Of Community Medicine And Public Health, 2017. **5**(1): p. 1-3.
6. Zaman, Q. and S. M, *Association between the Education and Thalassaemia: A Statistical Study*. Pakistan Journal of Statistics and Operation Research, 2006. **2**.
7. Leghari, B.F.a.I.U., *Social Determinants of Cousin Marriages (A Case Study of District Rawalpindi, Pakistan)*. FWU Journal of Social Sciences, 2020. **Vol. 14, No.2**, (Summer 2020): p. 155-163.
8. Oniya, O., et al., *A review of the reproductive consequences of consanguinity*. Eur J Obstet Gynecol Reprod Biol, 2019. **232**: p. 87-96.
9. Raiber, E., et al., *What Do Parents Want? Parental Spousal Preferences in China*. Economic Development and Cultural Change, 2022. **71**.
10. Fareed, M. and M. Afzal, *Genetics of consanguinity and inbreeding in health and disease*. Ann Hum Biol, 2017. **44**(2): p. 99-107.
11. Amuasi, G.K. and A. Eliasu, *Spousal Selection for Marriage by Tertiary Students in Wa Municipality of Ghana: Internal Factors in Perspective*. European Journal of Development Studies, 2022. **2**: p. 31-41.
12. Lima, S.O.A., et al., *A population-based study of inter-generational attitudes towards consanguineous marriages in north-eastern Brazil*. J Biosoc Sci, 2019. **51**(5): p. 683-697.
13. Zadran, S.K., M. Ilyas, and S. Dawari, *Genetic variants associated with diseases in Afghan population*. Mol Genet Genomic Med, 2021. **9**(5): p. e1608.

14. Chimah, O.U., et al., *Congenital malformations: Prevalence and characteristics of newborns admitted into Federal Medical Center, Asaba*. Health Sci Rep, 2022. **5**(3): p. e599.
15. Ciarleglio, L.J., et al., *Genetic counseling throughout the life cycle*. J Clin Invest, 2003. **112**(9): p. 1280-6.
16. Pameer, A., *Rate of Consanguineous Marriages in Afghan Society and Its Effects on Health*. International Journal of Health Sciences, 2023. **6**: p. 28-39.
17. Huits, R., et al., *Diagnosis and clinical relevance of co-inheritance of haemoglobin D-Punjab/ $\beta$ <sup>+</sup>-thalassemia traits in an immigrant Afghan family*. J Clin Pathol, 2022. **75**(12): p. 861-864.
18. Naibkhal, N. and E. Chitkara, *Consanguineous marriages increase risk of congenital anomalies-studies in four generation of an afghan family*. Biomedical Research, 2016. **27**: p. 34-39.
19. Bennett, R.L., et al., *Genetic Counseling and Screening of Consanguineous Couples and Their Offspring: Recommendations of the National Society of Genetic Counselors*. J Genet Couns, 2002. **11**(2): p. 97-119.
20. Akrami, S.M., *Genetics of consanguineous marriage: Impact and importance of counseling*. J Pediatr Genet, 2012. **1**(4): p. 217-20.