

Study of Genetic and Clinical Outcomes of Consanguineous Marriages

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Abstract

Consanguineous marriage (CM) or cousin marriage is a type of inter-familial union, defined as the marriage between two blood-related individuals who are second cousins or closer (inbreeding coefficient ≥ 0.0156). Marriage is one of the important vital event and entry in to reproductive life in India. Prevalence and pattern of marriage varies across the states, regions and according to their religion and caste. Consanguineous marriages have been practiced since the early existence of humans. Consanguinity is the marriage between close relatives. Though consanguineous matings have cultural and socioeconomic advantages, their offspring have an increased risk for recessive disorders. The risk is not uniform in all cases and it varies based on several factors. Consanguinity is a deeply rooted social trend among one-fifth of the world population mostly residing in the Middle East, West Asia and North Africa, as well as among emigrants from these communities now residing in North America, Europe and Australia. The mounting public awareness on prevention of congenital and genetic disorders in offspring is driving an increasing number of couples contemplating marriage and reproduction in highly consanguineous communities to seek counseling on consanguinity. The article discusses the different type of measures that can be taken by a consanguineous couple before and after marriage to avoid genetic disorders in their progeny.

Keywords: Consanguinity, Consanguineous marriages, Genetics, Disorders, Progeny, Recessive.

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INTRODUCTION

Consanguineous marriage, also known as cousin marriage, is when two people who are closely related by blood get married. In clinical genetics, this refers to marriages between people who are second cousins or closer. According to Bittles (1994), Consanguineous marriage (CM) or cousin marriage is a type of inter-familial union, defined as the marriage between two blood-related individuals who are second cousins or closer (inbreeding coefficient ≥ 0.0156). Over a billion of the global population live in communities where consanguineous marriage is a traditional and respected social trend of marital union (Saba *et al.*, 2021). The primary reasons for a preference for consanguineous marriage in communities with high consanguinity rates include maintaining the lineage solidarity of family, relative ease for the partners in finding a suitable spouse, backing the female status and improved relationships with in-laws, lowering the marital cost, enhancing the likelihood of getting better care for people in old age, and above all, better stability of marital relationship. These factors seem to carry more

significance in the context of protective and semi-protective cultures of many Middle Eastern, South and West Asian, and sub-Saharan African societies, which is why consanguineous marriage is most frequent in these countries (Bittles, 1994). If two individuals have a common ancestor, they are said to be consanguineous. A marriage between closely related individuals, up to second cousins is considered to be consanguineous marriage. Charles Darwin, Albert Einstein, Queen Victoria, Franklin Roosevelt etc. are some of the famous persons who married their cousins. Most human societies however primitive or geographically isolated, prohibit parent-offspring or brother-sister matings (marriage among first degree relatives) and this taboo is based on observation of abnormal children from such matings. Consanguineous unions are practiced among more than 1 billion of the world's population, however the amount of inbreeding varies in different populations. It is controlled by traditional and cultural practices. Though it is not allowed in some countries like China, Ethiopia, Philippines, Taiwan etc. It is high in North Africa, the Middle East and South Asia and is common in our country (Hamamy and Alwan, 2016).

It is estimated that one billion of the current global population live in communities with a preference for consanguineous marriage (Bittles and Black, 2010a; Modell and Darr, 2002). Consanguineous marriage is traditional and respected in most communities of North Africa, Middle East and West Asia, where intra-familial unions collectively account for 20–50+% of all marriages (Bittles, 2011; Hamamy *et al.*, 2011; Tadmouri *et al.*, 2009). In clinical genetics, a consanguineous marriage is defined as a union between two individuals who are related as second cousins or closer, with the inbreeding coefficient (F) equal or higher than 0.0156 (Bittles, 2001), where (F) represents a measure of the proportion of loci at which the offspring of a consanguineous union is expected to inherit identical gene copies from both parents. This includes unions termed first cousins, first cousins once removed and second cousins. In some communities, the highest inbreeding coefficients are reached with unions between double first cousins practiced among Arabs and uncle–niece marriages practiced in South India where (F) reaches 0.125 (Hamamy *et al.*, 2011).

Sharma *et al.*, 2020 in their studies reported that religion and north–south regional dichotomy in culture largely determine consanguineous marriage rather than socioeconomic condition in India. The overall prevalence of consanguineous marriage was 9.9%; the South region (23%) and North-East region (3.1%) showed the highest and lowest prevalences, respectively. Muslims had a higher prevalence (15%) than Hindus (9%). The prevalence of first cousin marriage (8.7%) was more than that of second cousin (0.7%) and of uncle–niece marriages (0.6%). Women living in urban areas and in nuclear families, having a higher level of education and belonging to affluent families were less likely to marry their cousins ($p < 0.01$). Women living in the South region of the country were more likely to marry their cousins, as well as uncles ($p < 0.001$). Close scrutiny of the trends in the results (odds ratios) revealed no clear relationship between socioeconomic condition and consanguineous marriage (Sharma *et al.*, 2020).

Consanguineous marriage is the legal union of male and female of a common ancestor related by blood. The most common prevailing form of consanguineous marriages is between first cousins. Middle East Asian countries and southern states in India show high prevalence. The highest percentage of consanguineous marriages is seen among the first cousins from both father's and mother's side, most commonly practice in the southern states of Andhra Pradesh, Telangana, Tamil Nadu, and Karnataka except Kerala. Importantly, the practice of consanguineous marriage is higher among the Muslims of North India and Hindus of Southern India, among the Other Backward Classes and the less educated population of the middle and richer wealth index (Acharya and Sahoo, 2021).

Bhasin and Nag (2017) showed that Ladakh muslim show increased fertility and decreased proportion of surviving children in consanguineous than in non-consanguineous marriage. They also reported that most prevalent type of consanguineous marriage is that between first cousins.

The actual reasons given for the preference of consanguineous marriages are primarily social. In communities with high consanguinity rates, sociological studies indicate that consanguineous marriage could enforce the couples' stability due to higher compatibility between husband and wife who share the same social relationships after marriage as before marriage, as well as the compatibility between the couple and other family members.

Consanguineous marriage may be more favourable for the women's status, including the wife's better relationship with her in-laws who could support her in time of need. There is a general belief that marrying within the family reduces the possibilities of hidden uncertainties in health and financial issues. It is believed that consanguinity strengthens family ties and enforces family solidarity, with cousin marriage providing excellent opportunities for the transmission of cultural values and cultural continuity (Sandridge *et al.*, 2010). Premarital negotiations regarding financial matters of marriage are more easily conducted and sometimes less costly. Wife's parents prefer to have their daughter living near them and to enjoy the presence of their grandchildren. Moreover, wealthy landlords may prefer to keep their property within the family (Bittles, 2001; Hamamy and Bittles, 2008).

PROS AND CONS

There is a great controversy on the benefits and risks of consanguineous marriages. The positive social effects of consanguineous marriages are strengthening kinship relations, stable marital relationship, financial advantage, cultural continuity, maintenance of family structure, political alliances etc. which are the reason for the preference for consanguineous marriages. The effect of consanguinity on normal human variations is not known. The negative health effects are increased incidence of rare genetic disorders, increase in mortality and morbidity. Though the effects of consanguinity on non-communicable diseases is still largely unexplored, a recent study revealed a significant increase in the incidence of common adult diseases like cancer, diabetes, hypertension etc. in consanguineous population (Bener and Mohammad, 2017).

We all carry more than 25,000 genes in each cell as pairs, usually a pair for each character (some characters like tallness can be governed by more than one pair of genes). Out of these, about 3-6 genes are deleterious, but as they are recessive or weak they do not manifest the disease. The reason is the other member of the pair is normal and dominant (heterozygous) and

suppresses the deleterious effect of the gene. The deleterious genes will be expressed only if there are two copies of them in an individual (homozygous recessive). Individuals with common ancestors are likely to share the same alleles and their progeny has an increased chance of being homozygous. Deleterious alleles are more frequently expressed in homozygotes, resulting in a variety of genetic disorders such as birth defects, mental retardation, deafness and blindness or reduced viability known as inbreeding depression. One of the consequences of consanguinity is an increase in frequency of homozygotes. Increase in homozygosity is applicable not only for abnormal alleles but also for normal alleles, i.e., consanguinity can also result in perfectly healthy offspring (Bittles *et al.*, 1992). For example, Cleopatra who is known for her wit, beauty and intelligence was the daughter of a brother and sister and great granddaughter of another brother and sister. The adverse genetic effects on health do not affect 90% of the offspring of consanguineous marriages (Hamamy, 2003).

Consanguinity risk is highest among families which carry severe autosomal recessive diseases. Though the risk of congenital anomalies among the newborns of first cousin matings is claimed to be double the frequency among general population, the genetic implications is not the same for all the cousins. The magnitude of risk depends on the frequency of defective genes in the population, the degree of relationship between the parents, whether there are repeated cousin marriages in the family etc. It may be more than double in certain populations basing on repeated cousin marriages through several generations. Sibs share 50% of their genes whereas uncle niece and first cousins share 25% and 12.5% respectively while second cousins share only 3% of their genes. In marriage between individuals beyond second cousins, the risk will be almost the same as that in general population. Hence there is no deleterious genetic effect on health in consanguinity beyond second cousins. The few deleterious recessive alleles present in any population, rarely achieve homozygosity if population is out-breeding. They are mostly passed on to future generations silently (hidden or masked state), that is in a single copy or heterozygous state and when the allele comes in contact with the same allele in the partner the genetic disease will surface again. Unrelated couple may also produce a child with serious birth defect if unfortunately both of them have defective alleles of the same gene which happens when the frequency of the recessive allele is high in the population. High levels of endogamy in the population can dramatically increase the possibility of homozygosity at any locus. That is the reason for the high frequency of rare autosomal recessive disorders in regions where consanguinity is high. According to one school of thought by avoiding consanguineous marriages, the gene will be carried on to the future generations silently without elimination. Some experts believe that inbreeding is helpful to a population by constantly

exposing harmful recessive genes to selection and thereby eliminating them from the population (Balwan and Saba, 2021; Balwan and Saba, 2020).

Genetics and Clinical outcomes of Consanguineous Marriages

Consanguineous marriage is customary in many societies, but leads to an increased birth prevalence of infants with severe recessive disorders. It is therefore often proposed that consanguineous marriage should be discouraged on medical grounds. However, several expert groups have pointed out that this proposal is inconsistent with the ethical principles of genetic counselling, overlooks the social importance of consanguineous marriage and is ineffective. Instead, they suggest that the custom increases the possibilities for effective genetic counselling, and recommend a concerted effort to identify families at increased risk, and to provide them with risk information and carrier testing when feasible (Modell and Darr, 2002).

The study by Bener *et al.*, 2007 showed that in a population with a high rate of consanguinity, there is a significant increase in the prevalence of common adult diseases like cancer, mental disorders, heart diseases, gastrointestinal disorders, hypertension and hearing deficit.

Turkey has a high rate of consanguineous marriages. Different nationwide surveys indicate that today 20-25% of marriages are consanguineous, with the rate having increased over the last 15 years. The results of many studies show that the rate of consanguinity among parents of children with rare recessive diseases is quite above Turkey's average and that the high consanguinity rate is one of the underlying factors of high infant and child mortality and fertility in Turkey (Tuncbilek, 2001).

The study carried out by Khoury and Massad (2000) showed that fertility, as measured by the number of pregnancies, taking into consideration marriage duration, was not affected by consanguinity. Twin pregnancies and abortions did not show any significant difference between consanguineous and non-consanguineous marriages. Consanguineous marriages showed significantly higher rates of still births and infant mortality in general. Within the consanguineous group, female infant mortality rates were significantly higher than those of males. Congenital malformations as reported by mothers of consanguineous marriages were significantly higher than those reported by mothers of non-consanguineous marriages.

Consanguineous marriages are more prevalent in the Muslim-majority state of Jammu and Kashmir than in other regions of India. According to a study conducted in the Bhimber district of Jammu and Kashmir, Pakistan, 62% of marriages in the area were consanguineous, with first-cousin unions being the most common type (Jabin

and Malik, 2014). The study also found that consanguinity was associated with ethnicity, family structure, language, and marriage arrangements. According to Jabin and Malik (2014), the current descriptive epidemiological study carried out in Bhimber district of Mirpur division of Jammu and Kashmir, Pakistan, demonstrated that consanguineous marriages were 62% of the total marriages ($F=0.0348$). First-cousin unions were the predominant type of marriages and constituted 50.13% of total marital unions. The estimates of inbreeding coefficient were higher in the literate subjects, and consanguinity was witnessed to be rising with increasing literacy level. Additionally, consanguinity was observed to be associated with ethnicity, family structure, language, and marriage arrangements. Based upon these data, a distinct sociobiological structure, with increased stratification and higher genomic homozygosity, is expected for this Kashmiri population. In this communication, we present detailed distribution of the types of marital unions and the incidences of consanguinity and inbreeding coefficient (F) across various sociodemographic strata of Bhimber/Mirpuri population. The results of this study would have implication not only for other endogamous populations of Pakistan but also for the sizeable Kashmiri community immigrated to Europe. In the Ladakh region of Jammu and Kashmir, consanguineous marriages are practiced by some Muslim population groups, but are not widely practiced among Buddhist Bodhs (Bhasin and Nag, 2002). According to Bhasin and Nag (2002), 503 married Muslim women belonging to Balti, Brokpa and Arghun population groups of Ladakh region have been studied. The incidence of consanguinity have been found relatively low at 19.3 percent, when compared with other Muslim population groups of northern India. The mean coefficient of inbreeding has been estimated as 0.0116 for Ladakh Muslims as a whole. The most prevalent type of consanguineous marriage is that between first cousins. The Ladakh Muslims also show increased fertility and decreased proportion of surviving children in consanguineous than in non-consanguineous marriage, as noticed elsewhere as well.

CONCLUSION

Consanguinity is a deeply rooted social trend with one billion people currently living in countries where consanguineous marriages are customary, and among them, one in every three marriages is between cousins. The rising public awareness on possible preventive measures for congenital disorders has led to an augmentation in the number of couples seeking preconception and premarital counseling on consanguinity. Inbreeding is a controversial subject and it is very difficult to run experiments to determine its possible effects in humans. It is not always harmful. It can produce perfectly normal offspring also provided there is no genetic disease running in the family or there is no earlier history of consanguinity. It is important to remember that random mating does not eliminate

deleterious genes from a population, but merely covers them up. While inbreeding does not create harmful genes, it merely tends to bring them out. It is wise to consider the possible genetic consequences before marriage itself, especially if there are any deleterious recessive genes running in a family. Premarital counseling is important as it causes awareness of the possible harmful consequences among the related young adults who are going to marry. Already married cousins can use preconception counseling services to maintain health and in case of couples with affected children, prenatal diagnosis can be done to avoid genetic disorders. Necessary marriage counseling centers to be established to impart the knowledge about marriage relation and genetic deformities to younger generation.

Conflict of Interest: The author declares that they have no conflict of interest.

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