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Genetic Counseling in Central Iran: Lack of Public Awareness about Genetic Diseases?

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Abstract: Objective: The prevalence of consanguineous marriage in Middle East Countries is higher than the rest of the word. In Iran, consanguineous marriages have a frequency between 15.9%- 47%, with the highest in Eastern provinces. To determine prevalence of consanguineous marriage, kind of referral, prevalence of genetic disorders, and reasons of the referral in couples referred to genetic centers in central Iran. Methods: 2141 couples referred from 2008 to 2013 to genetic centers in central Iran fill out a simple questionnaire designed for this study. Results: Our findings indicated that premarital, preconception, prenatal and postnatal counseling had the frequency of 46.8%, 33.9%, 9.6% and 9.6%. At least a genetic disease was diagnosed in 75.3% of the clients. Mental retardation has the highest frequency of 49% among the diseases in the pedigrees. Conclusion: The low frequency of the referrals for premarital counseling and high frequency of diseases in the couples' pedigree indicate the poor awareness of the residents in this part of Iran of the genetic centers services. There is a great need for planning educational programs to inform young people about the potential threats of consanguineous marriage and also the merging services in genetics for diagnosis of inherited diseases.

Keywords: Genetic counseling; Central Iran; Consanguinity.

1. Introduction

Consanguineous marriage is a marriage between two individuals related as second cousin or closer [1, 2]. An increased risk of autosomal recessive diseases is associated with the consanguineous marriage. The offspring of a consanguineous marriage is autozygous for 6.25% of all loci. Inbreeding coefficient (*F*) is a term to express the proportion of loci in an individual with identical copies of gene inherited from both parents. The mean inbreeding coefficient is calculated by the inbreeding coefficient of a specific category of consanguineous marriage and the proportion of this category in the population [3, 4]. The mean inbreeding coefficient and marriages between first cousins are used for comparison of consanguinity rates in different populations [5].

The prevalence of consanguineous marriage is different between and within populations. Ethnicity, religion, culture, and geography affect the rate of consanguinity. Approximately 20-25% of all marriages in North Africa, Middle East, West Asia and South India are consanguineous with the highest rate for first cousins. The consanguineous marriage in Arab countries is between 40-68% and in non-Arab countries such as Turkey and Iran 21.2% and 38%, respectively [6-8]. The main reasons given for the preference of consanguineous marriage are stability of marriage; strength of the unity of family members, maintenance of cultural values and property, ease of marital arrangements, better relations with in-laws, and health and financial advantages [9, 10].

In communities with high prevalence consanguineous marriage, misconception that cousin marriages on the paternal side of the family only result in genetic diseases limits the accuracy of risk assessed by Genetic counselors [11]. The recommendations for a couple with consanguineous marriage should be adjusted to the timing the couples

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are referred for counseling [12]. The aim of this study was to determine the prevalence of consanguinity marriage in referrals to genetic centers in central Iran, timing of referral, prevalence of genetic disorders in the family, and reasons of the referral.

2. Methods

This study was performed in a 5-year period from 2008 to 2013. Any couple referred for genetics counseling to participating counseling centers was requested to fill out a questionnaire. The total number of couples in the study was 2141 from the central of Iran. The questionnaire included demographic and specific questions related to age, level of education, relationship, and the presence of any inherited disease in the family. Data on types of diseases in the extended pedigree were recorded according to the clinical data presented by the referrals. The participants were categorized in four groups including couples referred for premarital counseling (PMC), preconception counseling (PCC), prenatal counseling (PNC), and postnatal counseling (PC) to determine the frequency of consanguinity in each group. Consanguineous marriages were classified into double first cousins, first cousins, second cousins and beyond second cousin by the degree of relationship between couples.

3. Results

Data of 2141 couples referred for genetic counseling at genetic centers, located in central Iran, indicated the frequency of referrals for each kinds of counseling. According to the data, premarital, preconception, prenatal and postnatal counseling had the frequency of 46.8%, 33.9%, 9.6% and 9.6% (Table 1). Among couples counseled at premarital and preconception stages 69.2% and 65.2% had consanguineous relationships, respectively. Patrilateral parallel cousins had the frequency of 62.1% among the couples with consanguineous relationship.

The genetic diseases observed in the pedigree of the couples were categorized in table 2. The diseases were recorded based on available clinical data. According to the table mental retardation was the most frequent abnormality in the pedigrees. The frequency of pedigrees with one and two members affected was 17.2% and 12.7%, respectively. Pedigrees with 5-10 and more than 11 members with MR have the frequency of 2.8% and 1.3%.

4. Discussion

Consanguineous marriage is highly practiced in nations in Middle East [11]. Consanguineous marriages in Iran ranges from 15.9%- 47%, with the lowest observed in the Northern provinces and the highest in the Eastern provinces [10]. Consanguinity increases the level of homozygosity, especially for recessive genes, and, thus, increases the proportion of genetic diseases with recessive inheritance [13].

In this study the rate of consanguinity and kind of counseling for the couples seeking counseling in genetic centers in central part of Iran were investigated. Our findings indicated that 46.8% of referrals were referred to the genetic counselors before marriage. From couples referred before marriage, 69.2% had consanguineous relationships, from which 55.1% were first cousins. Couples seeking counseling during PCC were the second group with highest percentage of referrals. 65.2% of these couples had consanguineous relationship of first, double or beyond second cousins. 33.1% of couples referred in PCC were first cousins. These data shows that despite the presence of genetic counseling centers for more than two decades in central part of Iran, the frequency of the referral couples who have consanguineous relationships were less than expected. It should be noticed Iran is among the countries with high consanguinity rate ranging from 15.9% to 47%. It is expected all the couples with consanguineous relationship be referred for counseling before marriage. However, according to this study approximately half of referred couples with first or double cousin relationships had never been referred for counseling before marriage. All the participants in this study have Islamic faith and are aware of importance counseling before marriage. However, because of their lack of awareness of the presence of genetic centers providing counseling, these couples were not exposed to counseling before marriage. Therefore, the main obstacles preventing couples from referring to genetic centers should be investigated precisely in order to offer genetic counseling services to couples from high risk families. In a similar study in genetic centers in southern provinces of Iran, authors indicated that the majority of clients were referred during PMC [14]. The highest frequency of premarital counseling in Southern Iran (80%) in contrast to our studied population (46%) warrants the need to seek for alternative approaches to educate public about the emerging advances in genetics for diagnosis and prevention of genetic diseases.

Among the couples referred before marriage, 30.8% were non relatives. These couples were referred because of the presence of genetic diseases, infertility, stillbirth and postnatal death in the extended pedigree. Despite the presence of a genetic disease in 75.3% of the clients, only 46.8% were referred to genetic counselors before marriage. 20% of the referrals had at least one repetitive genetic disorder in the pedigree. Among the diseases observed in the pedigrees, mental retardation (MR) was observed in 49.1% of the clients (at least in one member of the pedigree). And 17.2% and 12.7% of the pedigrees has one and two children with MR, respectively. The nearly close frequency of families with one and two children with MR indicates that few families with a child affected with MR benefited from genetic services. Proper genetic services when the first child was diagnosed with MR might have prevented the birth of additional members affected with MR in these families. In addition, among the referrals, 4.1% of the clients have more 5 members in their pedigree affected with MR. This also indicates the poor access of these families to genetic services for diagnosis and counseling. Therefore, Because of the burden that individuals with MR have in the society and their families, health professionals should identify families with multiple members to help

prevent the recurrence of additional members with the diseases. However, this might not be practical in the small communities where the family members hide their child disease and deny the existence of any member with similar disease. The Hearing loss and visual loss occurred in 15% and 7.1% of the studied pedigrees, respectively. Among the couples with a child affected with a genetic disease (10%), only 4% were referred when the child was alive. 6% of the parents were referred after their child death. The undiagnosed status of these dead children prevents precise estimation of recurrence risk of the disease in the family. Many of the families are not aware of the advances made in genetic diagnostics, and, because the initial diagnosis of their child disease was not informative, they will not refer to genetic centers for counseling. In recent years, next generation sequencing has helped physicians to find the genetic etiology of many diseases which had an undiagnosed status [15]. These advances are important for communities with high rate of consanguinity rate where the rare recessive genetic disorders occur in families.

5. Conclusion

Our finding indicates that genetic counseling is not well recognized among the residents in small villages and towns in central Iran. The high costs of genetic tests and low income of families living in these communities with high frequency of consanguineous marriage is another reason for couples' denial to refer to genetic centers. To address the reasons for consanguineous couples' denial to refer to genetic counselor, more standardized studies should be conducted in this part of the Iran.

Public media, institutions or religious communities should provide educational programs to increase the awareness about the risks of consanguineous marriage among young adults. These programs should target individuals at an early age of fifteen to be effective because in traditional families girls are engaged in late teen years in arranged marriages. Considering the increased rate of consanguineous marriages after 1979 in Iran [7, 16] planning nationwide programs to increase public awareness is necessary.

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Declaration of Interest

The authors declare that they have no conflict of interest.

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Legends

Table Legend

Table-1. Frequency of Kind of counseling in Central Iran
Table-2. Prevalence of genetic disorders in participants

Table-1. Frequency of Kind of counseling in Central Iran

Kind of counseling*	Relatives %			Non relatives %	Total %
	first cousins	double cousin	second cousin or beyond		
PMC	55.1	45.5	37.6	30.8	46.8
PCC	33.1	31.8	37.1	34.8	33.9
PNC	6.5	4.5	11.4	16.6	9.6
PC	5.3	18.2	13.9	17.9	9.6
Total	100	100	100	100	100

*PMC: premarital counseling, PCC: preconception counseling, PNC: prenatal counseling, PC postnatal counseling

Table- 2. Prevalence of genetic disorders in participants

	Disease Frequency (%)
Mental retardation	49
Congenital malformations	24.8
Neurological diseases	24.3
Hearing impairment	15
Blindness	7.1
Short stature	1.8
Hematological diseases	3.7
Others	19.1
Total	75.4