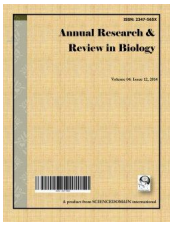

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Diverse Genetic Screening and Counseling throughout the Iranian Population

Zakieh Arab[1], Asghar Arshi[2], Mohammad-Javad Arshi[3], Zahra Yousefzadeh[1*] and Fatemeh Salmani[4]

[1]Genetic Counseling Center, Najafabad, Esfahan, Iran.
[2]Young Researchers and Elite Club, Islamic Azad University, Najafabad Branch, Najafabad, Iran.
[3]Department of Basic Science, Islamic Azad University, Shahrekord Branch, Shahrekord, Iran.
[4]Nursing and Midwifery Sciences Development Research Center, Najafabad Branch, Islamic Azad University, Najafabad, Iran.

Authors' contributions

This work was carried out in collaboration between all authors. All authors read and approved the final manuscript.

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ABSTRACT

Background and Aim: The recent decades have witnessed increasing possibilities for genetic testing and screening. In the Iran, since the 1970s, individuals and their family members could obtain genetic counselling for their own risk or diagnosis of a serious genetic disorder or that of their offspring. The aim of this study was to determine the results of screening for genetic disorders in Najafabad-Isfahan.
Methodology: This study was conducted on 2,500 families of Najafabad-Isfahan. 250 of these families were randomly referred for genetic counseling. In these families, the degree and type of disability, the death of children under 2 years of age, abortion and stillbirth were investigated and genetic counseling and tests were carried out in some families.
Results: The findings of this study showed family marriage have been 41% in students' parents and 31% in couples. The highest type of disability in order to frequency was consist of intellectual

**Corresponding author: E-mail: geneticspub@yahoo.com;*

disability, musculoskeletal, congenital disorders, deafness, microspheres, abortion and stillness, heart problems, vision, blood diseases, CP, death of children under 2 years of age, hydrocephalus, metabolic and autism.

Conclusion: Based on the results of present study it was shown that most disabilities happened in families with familial marriage or families with a history of disability and genetic counseling can be done by informing people at risk to lose the chance of having a child with a disability.

Keywords: Genetic counseling; screening; disability; Iran.

1. INTRODUCTION

Genetic screening is at the core of any proper prevention program, yet it is very recent, primitive, and almost nonexistent in Iran [1]. Genetic counselling is currently available in the United Kingdom (UK) for a variety of disorders and diseases with a substantial genetic component. Concerned with the cause, course, diagnosis and treatment of genetic disorders, genetic counselling is a medical specialty offering information-giving and psychosocial support to affected individuals and their families. Mirroring the broader purpose of genetic counselling [2], psychiatric genetic counselling helps individuals and their families with adaption to mental illness, by providing psychosocial support and an etiological information in the context of their own personal and family history. Rather than extending the 'technological paradigm' of current psychiatry, psychiatric genetic counselling has the potential to create a therapeutic context of empowerment and positive self-identity [3]. The historic and social structure of the Iranian population residing in Iran or elsewhere favors the high prevalence of a variety of rare and common genetic diseases. Perhaps, the most strongly associated reason would be the deeply rooted sociological norm of consanguinity, defined as a marriage among blood relatives, in addition to endogamous and arranged marriages within a community [4]. Genetic diseases in Iran, like other countries, are frequent with an incidence among the highest in the world [5]. Several factors contribute to this high incidence [6].

Today, psychiatric disorders are considered heritable conditions that affect individuals and their families' worldwide [7]. In the last two decades, genetic research has established the heritability and pathogenesis of psychiatric disorders as multifactorial and polygenic [8]. Genetic risk is thought to be the result of gene-gene and gene-environment interactions, which is likely to complicate the clinical interpretation of genetic causality [9]. Recent decades have witnessed increasing possibilities for genetic testing and screening. In clinical genetics, the doctor's office defined a secluded space for discussion of sensitive reproductive options in cases of elevated risk for genetic disorders in individuals or their offspring. When prenatal screening for all pregnant women became conceivable, the potential increase in scale made social and ethical concerns relevant for the whole of society. Whereas genetic testing in clinical genetic practice was widely accepted, prenatal screening at a population level met with unease [10].

According to the results of the census in 1996 in Iran, 3.9 percent of the households in the country have at least one disabled person (3.4 percent in urban areas and 4.7 percent in rural areas). About 10% of the world's population, about 600 million people, are affected by disabilities. But this is two times higher in Iran because of family marriages. According to studies conducted in Iran, more than 40% of disabilities are due to genetic disorders, and genetic diseases account for about 50% of childhood blindness and deafness and 60% of all severe intellectual disability [11]. Whenever new technological options, such as genetic tests, become available often political and public debates are called for to discuss the social and ethical ramifications [10]. Whereas reproduction is key to any society, balancing the tension between the interest of the individual and the collective regarding genetic reproductive issues is a delicate issue in modern democracies and a challenge for governmental policy making. The use or misuse of genetics by individuals or institutions in the first half of the previous century still sets the background of present-day arguments 2 [12].

Genetic counseling is the best way to recognize and prevent the birth of disabled children in families, and people according to cultural and ethnic topics tend to have family marriage. So the need for genetic screening before marriage in

this city is necessary. This study was performed to determine the effect of genetic disorders screening project in Najafabad-Isfahan city. The aim of this study was to determine the results of screening for genetic disorders in Najafabad-Isfahan.

2. MATERIALS AND METHODS

2.1 Sampling and Data Collection

Participants selected whether they would like to be involved in semi-structured interviews. Couples that their marriage was genetically in a high risk, referred to the genetic counseling center. In the other hand, students referred to the genetic counseling center after interview. In the genetic counseling center the questions of the study were based on biography, family tree, type of disability and inheritance. All data were collected between December 2016 and January 2017. Individuals who met the inclusion criteria were invited to participate in the study. The samples population included students (n = 1500) and couples (n = 1000). The number of 250 of these samples was randomly referred for genetic counseling. In these samples, the degree and type of disability, the death of children under 2 years of age, abortion and stillbirth were investigated and genetic counseling and tests were carried out in some samples. The experiment protocols were approved by the Studies Committee at the affiliated the Islamic Azad University, Shahrekord Branch, Shahrekord, Iran with 17621105 grant number.

2.2 Statistical Analyses

All data was analyzed using SAS Version 9.3 (SAS Institute, Inc., Carey, North Carolina). Participant responses regarding knowledge of genetic counseling/testing and risk appropriate screening intervals were compared overall and according to participant recruitment source using the Chi squared or Fisher's Exact test as appropriate. The level of statistical significance was set at *P*= .05.

3. RESULTS

In present study, 123 couples and 127 students in a total of 250 families of the total population interviewed had genetic disorders in one of the family members and genetic counseling was done for them (Table 1). Chi-square test showed that the frequency of family marriage was significantly higher in the parents of students (previous generation) than in new couples (new generation) (*P*= .03).

Table 1. Distribution of family genetic disorders in samples

Type of genetic disorder	Number	Percentage (%)
Intellectual disability	113	45.2
Musculoskeletal	71	28.4
Congenital disorders	33	13.2
Deafness	30	12
Death under 3 years of age	26	10.4
Abortion	19	7.6
Visual disturbances	18	7.2
Intellectual disability, Musculoskeletal	17	6.8
Congenital heart disorders	17	6.8
Microcephaly	16	6.4
Down syndrome	15	6
Blood disorders	12	4.8
Metabolic diseases	11	4.4
Hydrocephalus	10	4
CP	10	4
Autism	5	2
No genetic disorder	29	11.6

Also, according to the results of the Chi-square test, the distribution of the inheritance of genetic disorders in the studied families is not the same and the most frequent is autosomal recessive heredity (*P*= .001). The results shown in Fig. 1.

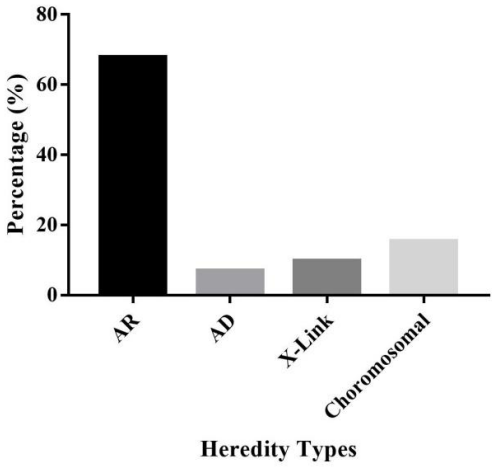

Fig. 1. Frequency distribution of the inheritance of disabilities in the investigated families

According to the studies, some of the disabilities were seen alone in the family and some of the disabilities were created at the same time in one person. See Table 2 for more information.

Table 2. Distribution of synchronization of disabilities in samples

Type of genetic disorder	The existence of disability		Disability with other disorders	
	Number	Percentage (%)	Number	Percentage (%)
Intellectual disability	27	23.9	86	76.1
Musculoskeletal	16	22.5	55	77.5
Congenital disorders	6	18.2	27	81.8
Deafness	7	23.3	23	76.7
Death under 3 years of age	8	30.7	18	69.3
Abortion	6	31.6	13	68.4
Visual disturbances	3	16.7	15	83.3
Heart disorders	5	29.4	12	70.6
Microcephaly	4	25	12	75
Blood disorders	3	25	9	75
Metabolic diseases	4	36.4	7	63.6
Hydrocephalus	2	20	8	80
CP	2	20	8	80
Autism	1	20	4	80

Fig. 2 indicates 40% of the followers who performed the diagnosis and were diagnosed were unable to perform the genetic test because they did not pay for the genetic tests or the lack of cooperation of the disabled person or other family members, 26.7% and 33.3% made a genetic test personal expense and welfare allowance respectively.

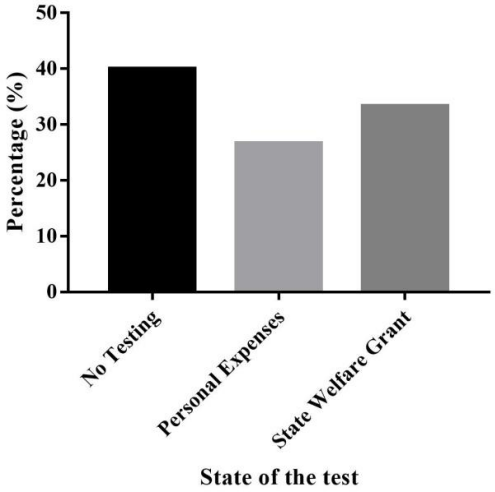

Fig. 2. Distribution of state of the test in the subjects examined

4. DISCUSSION

During the 1980s, it became increasingly clear that new techniques might enable mass screening of pregnant women. Maternal serum screening tests were developed to detect neural tube defects, and a few years later, Down syndrome, in a fetus. This potential increase of scale meant that discussions on reproductive options were no longer confined to the secluded space of a doctor–patient relationship, but that prenatal testing and screening had become relevant issues for the whole society [13]. In 1989, the Dutch government decided not to implement maternal serum screening for neural tube defects. The decision was based on the WHO criteria written by Wilson and Jungner (1968). The test characteristics were found to be inadequate; there were too many false positives and false negatives. Since there was no treatment available, the criterion that only treatable disorders should be screened was not met [14]. Anne Andermann in [15] indicated the relationship with the importance of genetic screening to reduce the mental pressure of families for childbirth, as well as reducing the amount of genetic disorders and also as a strategy for improving community health.

There is a strong preference for marrying a first cousin. The proportion of cousin marriage among urban families stayed constant. For all periods the proportion of cousin marriage among highly educated women was somewhat lower than among uneducated women. In a study Saadat et al. [16] reported that the overall rate of consanguineous marriage was 38.6% with a mean inbreeding coefficient (alpha) of 0.0185. First cousin marriages (27.9%) were the most common form of consanguineous union, with parallel patrilateral marriage especially favoured [16]. Several studies have been conducted on the subject of adultery marriages in Iran. Haji Sfandiari et al. [17] in Karaj, Iran conducted a study on 356 parents of exceptional children and 1,365 parents of normal children. The results of their study indicated 31.5% parents of exceptional children had married kinship while

the parents of ordinary children had 16.9% of kin marriages. In present study, which was conducted for the first time in Iran in the Najafabad as a pilot city, a plan for genetic disorders was ripening. Considering the population and number of families in this city, the percentage of genetic disorders in Najafabad was 4.5%. This statistic can be considered in contrast to the 2-4% birth rate of births of congenital disorders in the world and 2-3% of Intellectual disability in the world. In the present study, the most inheritance of disability in this city was estimated to be autosomal recessive, which could indicate that part of the reason for increasing the percentage of disability in this city is related to family marriages. In another study Shahri et al. investigated the prevalence of familial marriages in Ahwaz city located in south of Iran. The findings of this study showed a significant relationship between familial marriage and children's disability [18,19].

5. CONCLUSION

In order to prevent genetic disorders in other family members, the disease agent gene must first be diagnosed in a disabled person and then the defective gene examined in other family members. This fact suggests the need for more awareness for families to help diagnose the disease, as well as more cooperation from authorities to fund these trials. Also the results of present study recommend to involve and activate the family counselling in order to cope with believes and traditions which interpret the familial marriage in Iranian society. In the other hand, it recommend to involve and activate the school counselling in order to reconstruct believes related to familial marriage in Iranian society.

ETHICAL APPROVAL

This study was approved by institutional ethical committee and as per international standard or university standard; patient's written consent has been collected and preserved by the authors.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

REFERENCES

1. Nakouzi G, Kreidieh K, Yazbek S. A review of the diverse genetic disorders in the Lebanese population: Highlighting the urgency for community genetic services. J Community Genet. 2015;6(1):83-105.
2. Resta R, Biesecker BB, Bennett RL, Blum S, Estabrooks HS, Strecker MN, et al. A new definition of genetic counselling: National society of genetic counselors' task force report. J Genet Counsel. 2006;15(2):77-83.
3. Bracken P, Thomas P, Timimi S, Asen E, Behr G, Beuster C, et al. Psychiatry beyond the current paradigm. Br J Psychiatr. 2012;201(6):430-4.
4. Chouery E, Guissart C, Megarbane H, Aral B, Nassif C, Thauvin-Robinet C, Megarbane A. Craniosynostosis, anal anomalies, and porokeratosis (CDAGS syndrome): Case report and literature review. Eur J Med Genet. 2013;56(12): 674-7.
5. Tadmouri GO, Nair P, Obeid T, Al Ali MT, Al Khaja N, Hamamy HA. Consanguinity and reproductive health among Arabs. Reprod Health. 2009;6:17.
6. Lindner M, Abdoh G, Fang-Hoffmann J, Shabeck N, Al-Sayrafi M, AlJanahi M, Hoffmann GF. Implementation of extended neonatal screening and a metabolic unit in the State of Qatar: Developing and optimizing strategies in cooperation with the Neonatal Screening Center in Heidelberg. J Inherit Metab Dis. 2007;30(4):522-9.
7. Owen MJ, Cardno AG, O'Donovan MC. Psychiatric genetics: Back to the future. Mol Psychiatr. 2000;5:22-31.
8. Laegsgaard MM, Mors O. Psychiatric genetic testing: Attitudes and intentions among future users and providers. Am J Med Genet B. 2008;147B:375-84.
9. Finn CT, Smoller JW. Genetic counselling in psychiatry. Harv Rev Psychiatr. 2006;14:109-21.
10. Van ECG, Pieters T, Cornel M. Genetic screening and democracy: Lessons from debating genetic screening criteria in the Netherlands. J Community Genet. 2012;3(2):79-89.
11. Iossifov I, Zheng T, Baron M, Gilliam TC, Rzhetsky A. Genetic-linkage mapping of complex hereditary disorders to a whole-genome molecular-interaction network. Genome Res. 2008;18(7):1150-62.
12. Huijer M. Storytelling to enrich the democratic debate: The Dutch discussion on embryo selection for hereditary breast cancer. Bio Societies. 2009;4:223-38.

13. Van den Berg M, Timmermans DRM, Kleinveld JH, Garcia E, van Vugt JMG, van der Wal G. Accepting or declining the offer of prenatal screening for congenital defects: Test uptake and women's reasons. Prenat Diagn. 2005;25:84-90.
14. Wilson JMG, Jungner G. Principles and practice of screening for disease. WHO, Geneva; 1968.
15. Andermanna A. Genetic screening. Can Fam Physician. 2010;56:333-9.
16. Saadat M, Ansari-Lari M, Farhud DD. Consanguineous marriage in Iran. Ann Hum Biol. 2004;31(2):263-9.
17. Hajiesfandiary L, Afrouz Gh, Homan HA, Yaryari F. Comparative study of frequency of the six types of cross cousin and non-cousin marriage among parents of exceptional and normal children. Research on Exceptional Children. 2010;10(2):175-83.
18. Shahri P, Namadmalan M, Rafiee A, Haghighi Zadeh MH. A case-control study of prevalence of consanguineous marriage among parents of handicapped and healthy children in Ahvaz. Medical Research Journal of Ahvaz Jondishapur University of Medical Sciences. 2010;9(5):474-82.
19. Kakabaraee K, Seidy M. Comparison of frequency of cousin and non-cousin marriage among parents of disabled and normal children. Research and Health. 2012;2:162-7.

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