JLSB
Journal of Life Science and Biomedicine

J. Life Sci. Biomed. 4(5): 401-403, 2014

ISSN 2251-9939

The Impact of Genetic Counseling on Prevention of Mental Retardation

Pooneh Nikuei[1], Behnaz Mirzaei[2], Shide Rafaty[3], Fateme Poordarvish[1] and Rahman Mahdizadehdehosta [4*]
[1.] Molecular Medicine Research Center, Hormozgan University of Medical Sciences, Bandar Abbas, IRAN
[2.] Department of Genetics, Science and Research Branch Islamic Azad University, Tehran, Iran
[3.] Social Determinations in Health Promotion Research Center, Hormozgan University of Medical Sciences, Bandar Abbas, IRAN
[4.]Department of Biology, Bandar Jask Branch, Islamic Azad University, Bandar Jask, Iran

*Corresponding author's e-mail: r.mahdizadeh@iauba.ac.ir

ABSTRACT: Mental retardation has high prevalence in south of Iran because of consanguineous marriages. As no definite cure is available for mental retarded individuals, prevention is the best way .This study was designed to evaluate the impact of genetic counselling on prevention of mental retardation. This case control study was carried out between 2010-2013 and 120 women with mental retarded children were participated. All of them had another pregnancy after their mental retarded child. of those, 60 women had pregnancy after genetic counselling (case group) and 60 women had pregnancy without doing genetic counselling (control group).The study data was analysed using SPSS19.Genetic counselling before pregnancy was protective factor for having mental retarded child. (Odds Ratio 4.261, 95% confidence interval 1.312-13.834). Results indicated that 71.7% of parents in case group and 55% in control group had consanguineous marriages. Screening tests in pregnancy were done in 78.3% of mothers in case group and 21.7% in control group. Down syndrome was the most common cause of mental retardation in both groups. Genetic counselling is effective in prevention of mental retardation. It's suggested that health authorities inform and educate mothers about genetic counselling before pregnancy, specifically women with mental retarded children.
Keywords: Mental Retardation, Genetic Counselling, Pregnancy

ORIGINAL ARTICLE
Received 17 Jan. 2014
Accepted 18 Mar. 2014

INTRODUCTION

Mental retardation (MR) is one of the most important public health problems and the most common cause of referral to genetic centers [1]. It results to life-long harm effects for patients, families, and society and causes a psychological and economic expenditures. Mentally retarded patients have an intelligence quotient (IQ) below 70. Mild mental retardation with IQ between 50 and 70 (overall prevalence 1.5%) is more common than moderate and severe forms with IQ below 50 (prevalence 0.4%).

Mental retardation is characterized by impaired cognitive, linguistic, and social abilities. The incidence of mental retardation (MR) is 1-3% in the general population [2]. The etiology of MR is very heterogeneous and it can be caused by various genetic or environmental factors [3]. Causes of mental retardation could be genetic or non-genetic [4]. Up to 60% of cases, have no identifiable cause [5]. Genetic factors are the most common cause of severe MR [6]. Genetic causes classified as chromosomal abnormalities (aneuploidies, subtelomeric rearrangements, micro deletion or micro duplication syndrome), monogenic, metabolic, or multifactorial causes. High-resolution new techniques like -comparative genomic hybridization (CGH) arrays, and multiplex ligation probe amplification (MLPA) are useful tools to detect micro deletions and micro duplication all over the genome as causes of mental retardation. Genetic counseling is the process by which patients or relatives at risk of an inherited disorder are informed about the nature of the disorder, the role of genetics in their disorder and the options they have for management. Genetic counseling could reduce the impact of genetic disorders and aware families about prevention ways like prenatal diagnosis. The aim of this study was to investigate the impact of genetic counseling on prevention of mental retardation.

MATERIAL AND METHODS

Setting: The study conducted in Medical Genetic Counseling Center of Bandar Abbas in south of Iran with high consanguineous marriages with aim of impact of genetic counseling on mental retardation between 2010-2013 .

Subjects: This is a retrospective case control study. Participants were 120 women with at least one mental retarded children. All of them had another pregnancy after their mental retarded child .

Data collection: 60 women had their next pregnancy after referring to genetic counseling center (case group). Through genetic counseling complete family history was taken, pedigree was drawn, recurrence risk was estimated and prenatal screening tests were suggested for this group and 60 women in control group had their pregnancy without doing genetic counseling and were referred to Welfare organization centers for supportive cares. Case and

To cite this paper: Nikuei P., MirzaeiB. Rafaty SH., Poordarvish F. and MahdizadehdehostaR. 2014. The Impact of Genetic Counseling on Prevention of Mental Retardation. *J. Life Sci. Biomed.* 4(5):401-403.
Journal homepage:http://jlsb.science-line.com/

control groups were matched based on mothers and mental retarded children age. Mothers' age was below 40 years and mental retarded children age was below 14 years. A questionnaire was designed by researcher and variables like maternal age, number of mental retarded children, and age of children, history of genetic counseling before pregnancy, prenatal screening tests and result of karyotype were studied.

Data analysis: The study data was analyzed using software SPSS19.

RESULTS

Mean age of mothers in case group was 33 ± 4.9 and in control group was 34 ± 5.2. Mean age of mental retarded child was 7.8 ± 4.5 in case group and 10 ± 2.9 in control group. Mental retardation was 4 times more in case group (Odds Ratio 4.261 95% Confidence Interval 1.312 -13.834 (. In this study for 26.7% of mental retarded children in case group and 21% in control group karyotyping was done. Of those who did karyotype the most common cause of mental retardation in both groups were Down syndrome. (8.3% in case group and 11.3% in control group). Findings indicated that 71.7% of parents in case group and 55% in control group had consanguineous marriage. In case group 31.7% of mental retarded children were dead but all of mental retarded children in control group were alive. 23.3% of participants in case group and 26.7% of participants in control group had more than one mental retarded child. Moreover, 98.2% of mothers in case group and 100% of mothers in control group decided to do genetic counseling before their next pregnancy. After genetic counseling participants in case group (78.3 %) referred for doing non-invasive prenatal screening tests for trisomy's and neural tube defects during pregnancy. Of those 78.3 % participants, 25% were scheduled for amniocentesis because of abnormal first and second trimester screening tests to make sure the fetus was safe. Genetic counseling resulted in the diagnosis of 3.3% fetal abnormalities in women who did amniocentesis. They were referred for abortion because of abnormal results of amniocentesis.

Table 1. The frequency and percentage of how families in case group referred for genetic counseling

Referred from	Frequency	Percentage
Physicians	20	33.3
Friends	15	25.2
Advertising	15	25.2
Health centers	10	16.3

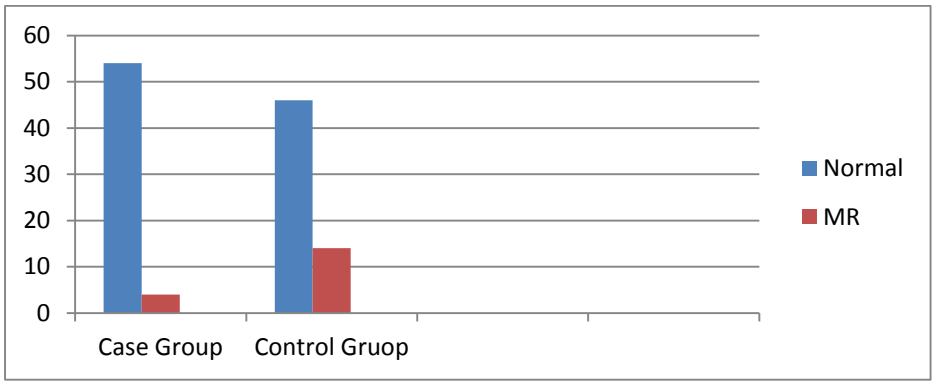

Fig1. Frequency of mental retarded and healthy children in case and control group

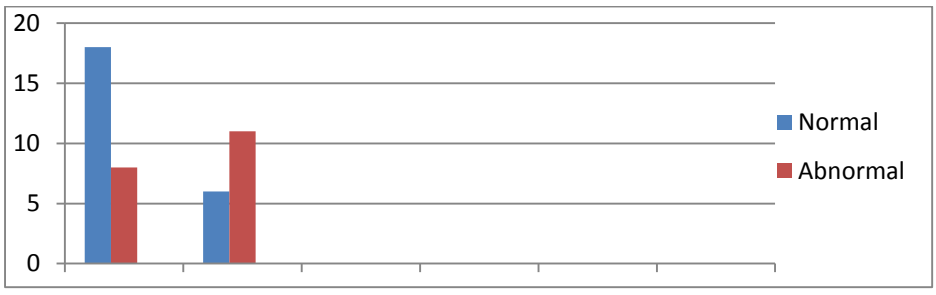

Fig2. Result of karyotype

DISCUSSION

To our knowledge this was one of the pioneering studies which investigated the effect of genetic counseling on prevention of mental retardation in families with at least one mental retarded child in Iran.

To cite this paper: Nikuei P., MirzaeiB. Rafaty SH., Poordarvish F. and MahdizadehdehostaR. 2014. The Impact of Genetic Counseling on Prevention of Mental Retardation. *J. Life Sci. Biomed.* 4(5):401-403.
Journal homepage:http://jlsb.science-line.com/

Down syndrome is the most common genetic cause for mental retardation [7, 8, and 9]. In our study the most cause of mental retardation in both groups were Down syndrome.

More than half of parents with mental retarded children had consanguineous marriage. In Madhavan study the risk of mental retardation in the offspring of consanguineously married was significantly higher [10]. In Saad study 61.5% of children with moderate and severe intellectual and developmental disability were offspring of parents who were biologically related, both first and second cousins[11] .

In our study in 78.3% of mothers in case group and 21.7% in control group noninvasive prenatal screening tests for trisomies and neural tube defects were done in pregnancy. In Kosec et.al study about Pregnant women's knowledge and attitudes to prenatal screening for fetal chromosomal abnormalities 73.1% of mothers had positive attitude toward first trimester combined screening tests and 16.9% had positive attitude toward second trimester biochemical screening [12].

Attention to genetic counseling before pregnancy could be an effective intervention for prevention of mental retardation in families with an affected child. In this study we found that genetic counseling before pregnancy is a protective factor for mental retardation that reduce recurrence risk 4 times so referring families with affected child to medical genetic counseling centers could be an effective way for prevention of this disability. Moreover because of high consanguinity marriages and autosomal recessive mental retardations due it, genetic counseling could plays an important role in diagnosis of the genetic diseases, carrier detection and prenatal diagnosis. So giving information and educating mothers about genetic counseling before pregnancy, specifically women with mental retarded children are recommended.

REFERENCES

1. Najmabadi, H., Motazacker, M.M., Garshasbi, M., Kahrizi ,K., Tzschach, A., Chen, W. et al. 2007.Homozygosity mapping in consanguineous families reveals extreme heterogeneity of non-syndromic autosomal recessive mental retardation and identifies 8 novel gene loci. Human genetics. 121(1):43-8.
2. Moser, H.W. 1995.A role for gene therapy in mental retardation. Mental Retardation and Developmental Disabilities Research Reviews, 1(1):4-6 .
3. Behjati, F., Firouzabadi, S.G., Kariminejad, R., Vameghi, R., Sajedi, F., Shafaghati, Y. ET al.2013 Genomic characterization of some Iranian children with idiopathic mental retardation using array comparative genomic hybridization. Indian Journal of Human Genetics. 19(4):443.
4. Tzschach, A. & Ropers, H. 2007.Genetics of mental retardation. Dtsch Arztebl; 104(20):1400-5 .
5. Rauch, A., Hoyer, J., Guth, S., Zweier, C., Kraus, C., Becker, C. et al. 2006.Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. American journal of medical genetics Part A. 140(19):2063-74 .
6. Ropers, HH. 2010. Genetics of early onset cognitive impairment. Annual review of genomics and human genetics. 11:161-87 .
7. Avent, N. 2013.Maternal plasma biomarkers for Down syndrome: present and future. Drugs of today 49(2):145-52 .
8. Comas, C., Torrents, M., Muñoz, A., Antolín, E., Figueras, F. & Echevarría, and M. 2002.Measurement of nuchal translucency as a single strategy in trisomy 21 screening: should we use any other marker? Obstetrics & Gynecology. 100(4):648-54 .
9. Haugen, O.H., Høvding, G. & Riise, R.2004. [Ocular changes in Down syndrome]. Tidsskrift for den Norske laegeforening: tidsskrift for praktisk medicin, NY raekke. 124(2):186-8 .
10. Madhavan, T. & Narayan, J. 1991.Consanguinity and mental retardation. Journal of Intellectual Disability Research. 35(2):133-9 .
11. Saad, H.A., Elbedour, S., Hallaq, E., Merrick, J. & Tenenbaum, A.2014. Consanguineous marriage and intellectual and developmental disabilities among Arab Bedouins children of the Negev region in Southern Israel: a pilot study. Frontiers in public health; 2 .
12. Košec, V., Zec, I., Tišlarić-Medenjak, D., Kuna, K., Šimundić, A.M., Lajtman-Križaić, M. ET al.2013.Pregnant Women's Knowledge and Attitudes to Prenatal Screening for Fetal Chromosomal Abnormalities: Croatian Multicentric Survey. Collegium antropologicum. 37(2):483-9.