Etiological characteristics of people with intellectual disability in Iran

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ABSTRACT

الأهداف: وصف الخصائص المسببة للإعاقة العقلية في أحد المراكز الإيرانية.

الطريقة: أُجريت هذه الدراسة المقطعية في مركز فاردافارد للرعاية الاجتماعية والتأهيل العقلي وذلك خلال الفترة من إبريل 2008م إلى سبتمبر 2008م حيث شملت 64 حالة مصابة بالتخلف العقلي. لقد تم تشخيص الحالات التي تعاني من التخلف العقلي أو تلك الحالات التي أظهرت تأخراً في علامات التطور العقلي، وقمنا كذلك بدراسة سجلات المرضى وعمل مقابلات مع الأهالي.

النتائج: لقد كان مجموع عدد الحالات 64 حالة مصابة بالتخلف العقلي وكان أكثرهم من الأطفال، ووصل عدد المرضى الذكور إلى 19 حالة (29.7%) فيما كان عدد الإناث 45 حالة (70.3%). لقد كان زواج الأقارب من الدرجة الأولى منتشراً في عائلات المرضى المصابين بالتخلف العقلي حيث كانت نسبة الإناث 48% ونسبة الذكور 52% من مجموع المرضى. وقد وصلت نسبة زواج الأقارب بين عائلات المرضى إلى 77% وكان نصفها تقريباً من زواج الأقارب من الدرجة الأولى. ووصلت نسبة الجهل بين عائلات المرضى المصابين بالتخلف العقلي إلى 34%. أشارت الدراسة بأن كلاً من: تعسر الولادة، ونقص وصول الأوكسجين إلى الجنين أثناء الولادة، وإصابة الأم بمرض مجموعي، وكذلك إصابة الجنين بعدوى من الأم أو عدوى بعد الولادة كلها تعد من العوامل الأساسية التي تؤدي إلى إصابة الجنين بالتخلف العقلي. بالإضافة إلى ذلك فقد كانت أكثر العائلات من الطبقات الاجتماعية الفقيرة.

خاتمة: تشير الدراسة بأن الإعاقات المعرفية مرتبطة بالعديد من العوامل، وكانت صلة القرابة من أكثر هذه العوامل خطورة، ولذلك فإن علينا لفت الانتباه إلى سلبيات زواج الأقارب الذي ينتشر بنسب عالية في الزواج التقليدي.

Objectives: To describe the etiological characteristics of intellectual disability in one Iranian center.

Methods: In a cross-sectional study, 64 cases with mental retardation (MR) were examined in the Intellectual Rehabilitation & Welfare Care Center of Vardavard, Iran between April and September 2008. Cases were diagnosed with MR or showed delay/regression of developmental milestones. We studied the records of patients and interviewed their families.

Results: A total of 64 cases were screened (most were children). The number of male patients was 19 (29.7%) and the females was 45 (70.3%). First degree relatives with mental retardation were found in the families of studied patients, among these relatives 48% were female and 52% were male. Up to 77% of the marriages were between relatives, approximately half between first cousins. The illiteracy rate reached 34% in the families of MR children. Hard labor, hypoxia during labor, mother's preexisting systemic disease, and maternal and neonatal infection were the most important factors for MR. Furthermore, most of the families were found to have low socioeconomic class.

Conclusion: Cognitive disabilities in children are multifactorial. Consanguinity was the main risk factor for MR and considering its high rate in our country due to traditional marriages, it should be modified.

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Mental retardation (MR) is a problem that involves approximately 1-3% of the general population. Cognitive disabilities in childhood are a major public health problem internationally, especially in developing countries.[1] Moreover, MR has a great influence on quality of life and productivity, which involves not only affected children but also their families and the society. In developing countries where risk factors for

childhood disabilities are not yet fully under control and the population-age pyramid is weighted toward the young group,[2] there is a great need to avoid these disabilities. Effective prevention, however, requires better information on risk factors and causes than is currently available for populations in the less developed world. This study reports the results of a primary investigation of risk factors for MR. The aim of this study was to describe etiological characteristics of people with intellectual disability in the fetal and childhood periods (prenatal, perinatal, neonatal, and postnatal) related to the prevalence of MR, and that may have implications for prevention.

Methods. This cross-sectional study was performed on 64 patients with MR at the Intellectual Rehabilitation & Welfare Care Center of Vardavard, Iran between April and September 2008. The Intellectual Rehabilitation & Welfare Care Center of Vardavard was established by the Government in 2000. All patients (3-40 years old) with MR living in this center were included in the study. Mental retardation was defined as significant deficits in cognitive function and adaptive behavior. The severity of MR was determined by Intelligence Quotient (IQ) level measured by the psychologist using a standard method. An IQ of 50-70 was considered mild, 35-49 moderate, and below 35 severe MR.[3] In our study, severe and profound cognitive disabilities (IQ under 25) were considered in a single category, called "serious". We also made 6 age classifications for these patients: <15 years, 16-20 years, 21-25 years, 26-30 years, 31-35 years and >36. The patients (children were accompanied by their parents), were examined carefully to detect phenotypic abnormalities and our examiners took their history. The genetic history was recorded in a prescribed format. Every noticeable point in chief complaints, birth, developmental, family and dietary history was noted. Since the patients were mentally retarded, their behavioral patterns were also noted carefully. Previous investigations were also recorded. For this study, 37 variables, for which data were available, were selected as potential risk factors for MR. The study protocol was approved by the Institutional Review Board of the Behzisti Organization and informed consent was obtained from the parents or caregivers.

Data analysis. All data including socio-demographic information was gathered from the family interviews. The screening, the care center, and the psychologic and medical examinations were recorded on forms and entered into a computerized database, and linked by identification numbers. Accuracy checks, and necessary corrections were made before and after the data were entered into the database. Data analysis was carried out using the Statistical Package for Social Sciences (SPSS Inc, Chicago, IL, USA) software version 15.00 for Windows and descriptive statistics were applied, including frequency of each variable and its distribution.

Results. Out of 64 cases included, most patients were in the age category of 16-20 years (n=23, 36%), patients' age ranged from 3-40 years. There were 19 (29.7%) male patients and 45 (70.3%) female patients. First degree relatives with MR were found in the families of studied patients, among these relatives, 48% were female and 52% were male. The whole family in 61 of 64 cases were available and studied, and it was found that there were at least 4 other relatives with cognitive disability besides the studied case in each family, which was estimated up to 136 (55%) for male and 110 (45%) for female relatives. The high rate of consanguinity played an important role in MR. Up to 77% of the marriages were between relatives, approximately half between first cousins. Forty-one families (64%) had more than one MR child and the interval between the first and second offspring was at least 5-6 years. Each family had a mean of 5 children, and the child death rate in each family was one out of 5 (20%). The IQ was less than 25 in 95% (severe MR) of children below 15 years. The illiteracy rate reached 34% in the families of MR children, while the primary school literacy level was found in 33%. Only 5% of the families showed academic studies at the bachelor degree level. Of 64 cases with MR, 15 cases (23%) had microcephalia, 15 (23%) had macrocephalia, and the rest 34 (54%) were normocephalic. Perinatal malformations such as hypoxic encephalopathy were observed in 18 mothers, which comprised the majority in the mothers' birth history. Dystocia (hard and prolonged delivery) followed perinatal malformations, and maternal history of diseases such as epilepsia, goiter, psychologic disorders, and diabetes followed dystocia. Pre and perinatal infection (neonatal sepsis especially gram negative type, TORCH infection, vaginal and urethral infections) were the other causes of MR.

A definite reason for MR attributed to a metabolic, autosomal, or gender-chromosome linked disease was only observed in 19% of cases, and there was no attributable disease in other cases with MR. Also, it was found that 77% families with at least one child with cognitive disabilities had a lower than normal socioeconomic state, and the remaining 23% could afford to live a normal life. In 17% of female, and 64% of male cases with MR, a lack of secondary puberty signs was noted. **Table 1** demonstrates detailed findings of the studied patients.

Discussion. Mental retardation is a serious burden for families and society. The results of this study are

Table 1 - Screened patients and frequencies of determinant risk factors for mental retardation (MR).

Variable	n	(%)
Etiology for MR		
Monogenic	45	(70)
Chromosomal	2	(3)
Environmental	12	(19)
Multifactorial	5	(8)
Risk factors for MR		
Mutation of a dominant gene	7	(11)
Consanguinity (recessive gene)	32	(50)
Gender chromosome related	5	(8)
Mother's high age/chromosomal	6	(9.5)
Multifactorial	14	(21.5)
Recurrence risk for MR		
Up to 1%	4	(6.3)
2-4%	2	(3.1)
5%	14	(21.8)
25%	36	(53.3)
50%	8	(12.5)
Potential risk factors with recurrent cases of MR		
Mental retardation	16	(25)
Skull anomalies/skeletal malformation	9	(14)
Paralysis	6	(9.5)
Epilepsy	3	(4.7)
Other	30	(46.8)
Cranial nerve examination		
Intact	22	(34.5)
With some degree of disability	42	(65.5)
History of epilepsy		
Epileptic	22	(34)
Normal	42	(66)
History of hyperactivity		
Hyperactive	30	(47)
Normal	37	(53)
History of neuromuscular and skeletal disorders		
Hypotonia	21	(33)
Neuromuscular disorders	14	(22)
Deformities	17	(27)
Other	12	(18)
Malformations in physical examination		
Ears	30	(46.8)
Eyes	24	(37.5)
Nose	12	(18.7)
Mouth	23	(50)
Cardiopulmonary	5	(7.8)
Speech disorders		
Normal speech	24	(37.5)
Speech disabilities	40	(62.5)
Birth and prenatal history		
Hypoxic encephalopathy	18	(28)
Dystocia	16	(25)
Mother's underlying diseases	15	(23.5)
Maternal infection	12	(19)
Other	3	(4.5)

in agreement with those of pilot studies conducted in developing countries.[4-6] In our study, the number of MR girls was higher than the number of MR boys, showing that families tend to keep the boys at home and use them for simple works. There are also negative cultural and social aspects that make the family send MR girls to the care center, which is in contrast to the findings on MR children in Kenya.[7] We found consanguinity (defined here to include offspring of parents related as first cousins or uncle-niece or second) to be responsible for 77% of MR in the studied patients. Other studies conducted in similar regions have also supported this fact.[3,8] We also found that parental illiteracy and low level of knowledge, especially maternal illiteracy, were one of the factors strongly affecting MR.[9,10] In the present survey, genetic factors were the most frequent etiology for MR in children (70%).

Regarding pre, peri, and post natal factors responsible for MR, some factors were remarkably attributed to the prevalence of MR including hard and prolonged labor, hypoxic encephalopathy during delivery, maternal underlying history of goiter, seizure, diabetes, and psychologic disorders, history of maternal infection or fever during pregnancy, lack of prenatal care, and postnatal sepsis and high maternal age.[11-16] The majority of MR children (60/64, 93%) had coexisting motor, seizure, vision, and/or hearing and speech disabilities. Among children with MR, a specific cause for the disability was identified in only 8% with a diagnosis of metabolic disease; these included 4 cases of phenylketonuria, and one case of glutaric aciduria Type I attributed to postnatal brain infections. Five cases were diagnosed with Down Syndrome and in other cases there were not enough data in their records. In these children, no biochemical assessments were ever performed.

No control group, the descriptive nature of study, and no genetic analysis are the main limitations of present study.

In conclusion, intellectual disabilities, especially in children, have multifactorial etiology. However, in our study, consanguinity and genetic factors were the main risk factors for MR. Considering the high traditional prevalence of consanguinity in our country, these marriages should be modified to avoid such events, or in cases of consanguineous marriages preventive measures such as prenatal diagnosis or preimplantation genetic diagnosis should be applied to reduce genetic intellectual disabilities. Although our screening program revealed some aspects of the etiologic characteristics of MR, we believe this study is still in the primitive phase, and further surveys on a larger scale are required.

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