Prevalence of Congenital Malformations

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Abstract- Congenital malformation (CM) will begin to emerge as one of the major childhood health problems .Treatment and rehabilitation of children with congenital malformations are costly and complete recovery is usually impossible. The aim of this study was to determine frequency of CM in Yazd central city of the Islamic Republic of Iran to find out if there has been any difference in the rate and types of CM in this area. This descriptive-observational study carried on 4800 births delivered at all maternity hospitals in Yazd from October 2003 to June 2004. Prevalence of CM was 2.83% (2.86 % in male and 2.68 % in female) out of the 136 cases 69(51.88%) were males and 64 (48.12%) were females and 3 with ambiguous genitalia. Positive family history of CM in sibling was in only 6 cases (4.41%).Overall, musculoskeletal (0.83%), central nervous system (0.47%) and genital system (0.37%) were accounted as the most common. Frequency of CM was more seen in still birth (12.5%) as in comparison to live birth (2.71%). There was not statistical difference between prevalence of CM and neonatal's gender, gestational age, birth order and mother's age, drug ingestion, illness and parental consanguinity. In this study the overall prevalence of congenital malformation among the newborn was higher than those previous reported in Iran and determining the causes of this difference needs more extensive studies.

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Key words: Congenital malformation, stillbirth, musculoskelet anomaly , CNS anomaly, cleft lip and palate

Introduction

Congenital malformation (CM) will begin to emerge as one of the major childhood health problems.Treatment and rehabilitation of children with congenital malformations is costly and complete recovery is usually impossible. CM can be separated into those that represent a single primary defect in development and those that represent a multiple malformation syndrome. For most of single primary defect the etiology is unknown, however most are explained based on multi-factorial inheritance. The etiology of malformation can be divided into genetic (multifactorial, Single gene, or chromosomal), environmental factors and teratogenic agents [maternal condition (alcoholism, diabetes, endocrinopathy PKU, nutritional deficiency), infections, mechanical problems, chemicals agents, drugs, radiation, hyperthermia, etc.] and unknown. Approximately, 66% of major malformations have no recognized etiology and most of them have multifactorial inheritance (1-3). Considerable variations in the frequency of congenital malformations in different populations have been reported, from 4.3% in Taiwan(4) to 7.92% in the united Arab Emirates (5) 2.46% and in Oman (6). Surveys on congenital malformations in Iran have been carried out in Tehran 3.5% and 2.3%, (6,7) Arak 1.04% (8) and Gorgan 1.01 (9) there may be regional variations in the pattern of CM. A similar study has not been conducted among newborns in yazd the present study was carried out to record the pattern of CM in this area a central city of Iran.

Patients and Methods

This was a descriptive, cross-sectional study of newborns and stillborn babies delivered at all maternity hospitals in yazd during 8 month period, from October 2003 to June 2004.

All live and stillborn newborns delivered in these hospitals during the investigation were examined and screened for congenital malformations by pediatricians. The medical records of newborns with congenital mal-

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formations were subsequently extracted for detailed study. Data collection was performed by means of structured from which contained two parts, similar to another study (7). At first part, variables recorded were about maternal characters and included the date of admission, age, history of chronic illness, drug ingestion, exposure to X-ray, history of CM in other offspring, parental consanguinity, and were obtained by interviewing with neonates' mother. The second part was about neonatal characters including live, or stillbirth, gestational age, birth order, sex, existence of CM and type of it, which were collected from medical records. No autopsy examinations were performed. The type of birth defects were classified by the diagnostic standardization of congenital malformation from the International classification of diseases (ICD-10) codes.Tables format is the same as that of Gorgan study (9) .Data were analyzed by SPSS 13. The rates of malformed newborns and malformations were compared with statistical t-test and chi-squared tests. The level of significance was P <0.05.

Results

During the 8 month period, 4800 newborns were delivered among whom 2411 were males and 2386 females and 3 with ambiguous genitalia. Out of these, 136 newborns were diagnosed with congenital malformations. Prevalence CM in this sample was 2.83% (69 males, 64 females, 3 with ambiguous genitalia). Sex distribution of CM shows in Table 1. There was not statistical difference between the rate of CM and newborn's gender (P = 0.1). In this study total stillbirth among the 4800 newborn was 56 (1.16%). The frequency of CM in stillbirth and live birth shows in Table 2 P<0.05, and statistical difference was found between frequency of CM in stillbirth and in live birth.

Table 3 shows the ICD-10 classification of the different types of congenital malformation. Some newborns had a multiplicity of malformations, so that the total number of congenital malformations exceeded the number of affected newborns.

Table 1. Prevalence of congenital malformation (CM) by sex in Yazd

Sex	No newborns delivered	No with CA	Total percentage
Male	2411	69	2.86
Female	2386	64	2.68
Ambiguous genitalia	3	3	100
All birth	4800	1 36	2.83

P. value= 0.1

Table 2. Frequency of congenital malformation (CM) in live birth and Stillbirth in Yazd

No. of newborns delivered	Total	No. of newborns with CM	Total percentage of CM
Live birth	4744	129	2.71
Stillbirth	56	7	12.56
All birth	4800	136	2.83

P. value<0.05

Table 3. All birth with CM by system according to the international Classification of Disease (ICD-10) in Yazd

Malformation system	No of CM	CM percentage (from220)
Muscloskeletal system	73	
Polydactyly	21	9.54
Clubfoots	19	8.63
CDH	9	4.09
Syndactyly	9	4.09
Brachydactyly	7	3.18
Clubhand	5	2.27
Rizomelia	3	1.36
Central nervous system	26	
Hydrocephaly	9	4.09
Meningomyelocele	7	3.18
Anencephaly	6	2.72
Microcephaly	4	1.81
Genitourinary system	25	
Hypospadias	14	6.36
Undescended testicle	7	3.18
Ambiguos genitalia	3	1.36
Epispadias	1	0.45
Eye,ear,face and neck	24	
Hypertelorism	10	4.54
Low set ears	4	1.81
Abnormal pinna	3	1.36
Microgenathism	2	0.9
Anophthlmos	2	0.9
Microphthalmos	1	0.45
Cataract	1	0.45
CA of nose ,unspecified	1	0.45
Cleft lip and Cleft palate	21	
Cleft lip with Cleft palate	4	1.81
Cleft lip	9	4.09
Cleft palate	8	3.63
Digestive system	21	
Imperforate anus	6	2.72
Atresia of esophagus with TE fistula	5	2.27
	4	1.81
High arched palate	4	1.81
Omphalocele	2	0.9
Gastroschisis	17	
Cardiovascular	9	7.72
Chromosomal abnormality	9	4.09
Down's syndrome	4	
Respiratory system	4	1.81
Choanal atresia		
Total	220	100

Table 4. Frequency of congenital malformation (CM)

Location / reference	CM Rate per 1000 birth	
	Live birth	All birth
Yazd, Iran	27.95	28.33
Tehran, Iran (6)	35	-
Tehran, Iran (7)	24.1	-
Arak, Iran(8)	10.4	-
Gorgan, Iran (9)	10.1	-
Oman (14)	-	24.6
Bahrain(10)	27	-
Arab Emarates(4)	7.89	7.92
Beirut, Lebanon(13)	-	16.5
Maharashtra , India (12)	10.8	12.8

Altogether 220 anomalies were documented in136 newborns. The musculoskeletal system was the most affected, involving 40 out of 136 patients (29.41%). Among this group, the most frequent anomalies were polydactyly, clubfoot, and CDH respectively. Anomalies of the central nervous system were second in frequency which involved 23 out of 136 patients (16.91%), and in this group the most common anomaly was hydrocephaly. Genitourinary system involved 18 out of 136 patients (13.23 %), and most common malformation was hypospadias. Anomalies of the eyes, ears and neck involved 17 out of 136 patients (12.5%), and in this group the most common anomaly was low set ear. Digestive system problems involved 15 patients (11.02%), among this group imperforate anus was the most common malformation detected. Circulatory system involved 12 patients (8.82 %). Cleft palate with or without cleft lip was seen in 11 patients (8.08%).

Discussions

In this study, the overall prevalence of congenital malformation among the newborn was 2.83% that is near to reports from Bahrain (2.7%) (10) and is lower than of Tehran (3.5%) (6), India(3.6%) (11), and is higher than those of previous reports from Iran, Tehran (2.4%), Arak (1.04%), and Gorgan (1.01) (7-9), and from India (1.28%) (12), United Arab Emarat (0.79%) (4) (Table. 4). These variations between different studies could be explained by the effect of different racial, ethnic and social factors in various parts of the world or different geographical, and socioeconomic factors. Similar study have not been previously performed in this city and higher frequency in Yazd may be due to industrial pollution , environmental and chemical factors, nutritional status and habits, high consanguinity marriage, etc. Other explanations for these variations in birth defect prevalence are the type of sample and the criteria for diagnosis. For determining the causes of this difference need in designing more extensive study.

Table.5 shows the frequency and comparison of difference types of congenital malformations in Yazd with other studies in Iran. In this study the rate of malformations in male newborns is nearly to that of females, and we did not observe any sex predilection of malformation and it is different from reports of Arak(8) Gorgan(9) that male newborns were more affected than females. In the present study, the commonest system involved was the musculoskeletal system, which agrees with reports from other parts of Iran (6-9) and other countries(10,12,13) but in Oman (14) , libyan Jamahiriya(15), and in the United Arab Emirates (4) CNS and GI, and Cardiovascular system anomalies were the most common respectively. In our study the most common musculoskeletal anomaly was polydactyly (4.37 per 1000 all births) which is nearly ten times of the reported from Tehran (0.44 per1000) (7), but CDH was the most common musculoskeletal anomaly reported from other parts of Iran (7-9). The rate of clubfoot was 3.95 per1000 which is higher than reported from other parts of this country and Denmark (16), Sweden (17). In this study the most common CNS malformation was hydrocephaly the same as one report from Tehran (7), the rate of hydrocephaly was 1.87per 1000. But in reported from Gorgan (9) the most common malformation of the CNS was meningomyelocele.

Table 5. Comparison of different type of congenital malformation (CM) in Yazd with other studies (No. of malformation system per 1000 birth)

location	Clubfoot	Anencephaly	Cystic spina bifida	Cleft palate with/without cleft lip	Imperforate anus	Hypospadias	Down syndrome
Yazd present study	3.95	1.25	1.45	4.37	1.25	2.91	1.87
Tehran(6)	2.9	0.08	0.92	1.61	0.38	4.45	1.23
Gorgan (9)	1.5	0.8	1.8	1.4	1.3	1.8	0.6
Arak (8)	1.4	0.4	1.2	1.6	-	-	-
Tehran(7)	0.43	1.3	0.87	2.18	-	0.43	0.43

In present study rate of cleft palate with or without cleft lip was 4.37per 1000 that is higher than reports from Tehran (2.18 per 1000) (7), Gorgan (1.4 per 1000) (9), Shiraz (0.8 per 1000) (18), and reports from other countries (19-23).

The most common genitourinary malformation was hypospadias which is the same as what reported from Gorgan (9) and Tehran (7), the rate of hypospadias was 2.91 per 1000 that higher than more reports from Iran (7,9), but lower than Singapore (4.12 per 1000) (24). The rate of imperforate anus was 1.25 per 1000 which is lower than in Gorgan (1.3 per 1000) and higher than in Denmark (0.38 per 1000) (25). In our study, the rate of Down syndrome was 1.87 per 1000 which is higher than reported from Tehran (6,7), India (12), and lower than that of Galway (26).

Congenital malformations were seen more significantly in still births as compared to live birth which is the same as reported from Gorgan (9) and India (12).

The difference between the frequency of types of congenital malformation in different parts of this country and reports from other countries may be due not only to genetic background but also to geographic nutritional and socioeconomic differences. More research is needed to determine the factors underlying the various types of congenital malformation encountered in this area.

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