Arch Iranian Med 2008; 11 (2): 148 – 151

Original Article

The Prevalence at Birth of Overt Congenital Anomalies in Urmia, Northwestern Iran

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Background: **Congenital anomalies play a significant role in perinatal and infantile morbidity and mortality. There is a variation in the frequency of congenital anomalies in different populations. Determination of the prevalence of different types of congenital anomalies may help plan primary prevention measures for these anomalies.**

The objective of this study was to determine the prevalence at birth of overt congenital anomalies in Urmia, in the northwest of Iran.

Methods: **In a cross-sectional hospital-based study, charts of 14,121 deliveries including live-births and stillbirths during the period from January 2001 through June 2005 were studied.**

Results: **A total of 264 (187 per 10,000 births) anomalies were detected. The anomaly categories with the highest prevalence were nervous system defects (52.65%) followed by musculoskeletal defects (23.86%). The total prevalence at birth of overt congenital anomalies was 1.87%. The rates for live- and stillbirths were 1.17% and 40.7%, respectively.**

Conclusion: **Considering the high frequency of central nervous system anomalies recorded in our study, it seems to be reasonable to pay more attention to the role of periconception vitamin supplementation for the primary prevention of congenital anomalies, particularly neural tube defects.**

Archives of Iranian Medicine, Volume 11, Number 2, 2008: 148 – 151.

Keywords: Birth defect • congenital anomaly • prevalence

Introduction

Major congenital anomalies occur in approximately 2 - 3% of births with a variable frequency in different populations ranging from 1.07% in Japan[1] to 4.3% in Taiwan[2]. Congenital anomalies contribute a significant proportion of perinatal and infant morbidity and mortality. Structural anomalies are considered overt when they are visible on inspection, otherwise they are considered "occult". Considering the elimination or control of some infectious diseases, congenital anomalies are increasingly playing a major role in the mortality and morbidity of children.[3,4] On the other hand, treatment and rehabilitation of these morbid children is difficult and costly.[4,5] Finding the variation in the frequency of congenital anomalies in different parts of the country may be helpful for health system authorities in planning healthcare measures for possible prevention of such anomalies.

Studies on the prevalence of congenital anomalies have already been carried out in some cities of Iran including Arak,[6] Hamadan,[7] Tehran,[8-10] and more recently, in Gorgan.[11] The present study was carried out to determine the prevalence at birth of overt congenital anomalies in Urmia, northwest of Iran, which to the best of our knowledge, is the first one of its kind in West Azerbaijan Province, Iran.

Materials and Methods

We designed a cross-sectional observational

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Accepted for publication: 19 September 2007

study to survey birth records over a 4.5-year period, from January 2001 through June 2005, at Motahhari Hospital which is a referral maternity hospital in West Azerbaijan, a province in northwest of Iran with a population of 2.9 million. Data were collected from documented hospital-based records of 14,121 charts including live-births and stillbirths. The documented information in the charts included the type of congenital anomalies in the affected cases, gender and weight of the fetus or neonate, maternal age, type of delivery, and gestational age. The main outcome measures were the total prevalence rate of overt congenital anomalies, the rate among live-born, and the rate among stillborn cases. The χ^2 test was used to test differences between the prevalence rates.

Results

During the 4.5-year study period, 264 overt congenital anomalies were detected among 14,121 live- and stillbirths, yielding a prevalence of 1.87% [95% confidence interval (CI): 1.65% to 2.09%). Sixty-one cases with minor nonspecified anomalies and 11 with hydrops fetalis were excluded from the study. The total prevalence rate of congenital anomalies for the period from 2001 through 2005 was 187/10,000; 1.17% for live- births and 40.7% for stillbirths. The types of anomalies were classified according to the international classification of diseases codes, version 10 (ICD-10) (Table 1). The rates of congenital anomalies were calculated per 10,000 total births including live- and stillbirths.

Table 1. Classification of congenital anomalies according to the international classification of diseases codes; version 10 (ICD-10).

Type of anomalies	ICD-10	No. of Cases	% among all anomalies	Per 10,000 births
Central nervous system	Q00-Q07	139	52.65	98.4
Anencephaly	Q00	78	29.55	55.2
Encephalocele	Q01	4	1.52	2.8
Spina bifida	Q05	35	13.26	24.8
Hydrocephaly	Q03	21	7.95	14.9
Microcephaly	Q02	1	0.38	0.7
Musculoskeletal system	Q65-Q79	63	23.86	44.6
Clubfoot	Q66	27	10.23	19.1
Amelia	Q71.0	1	0.38	0.7
Meromelia	Q71.8	11	4.17	7.8
Polydactyly	Q69	16	6.06	11.3
Syndactyly	Q70	1	0.38	0.7
Gastroschisis	Q79.3	4	1.52	2.8
Exomphalos	Q79.2	3	1.14	2.1
Digestive system	Q35-Q45	19	7.20	13.5
Cleft lip	Q36	8	3.03	5.7
Cleft palate	Q35	2	0.76	1.4
Clef lip with cleft palate	Q37	6	2.27	4.2
Imperforated anus	Q42.3	3	1.14	2.1
Urogenital system	Q50-Q64	18	6.82	12.7
Hypospadias	Q54	11	4.17	7.8
Epispadias	Q64.0	2	0.76	1.4
Ambiguous genitalia	Q56	5	1.89	3.5
Ear and neck	Q16-Q18	11	4.17	7.8
Ear anomalies	Q17	6	2.27	4.2
Branchial cyst/cleft	Q18.0	4	1.52	2.8
Anotia	Q17.8	1	0.38	0.7
Chromosomal anomalies	Q90-Q99	13	4.92	9.2
Down syndrome	Q90	13	4.92	9.2
Other anomalies	Q89	1	0.38	0.7
Conjoined twins	Q89.4	1	0.38	0.7
Total		**264**	**100**	**187**

Considering the categories of anomalies, based on the ICD-10 classification, the central nervous system was the most affected system followed by the musculoskeletal system.

To find any possible correlations between the frequency of congenital anomalies and gender of the newborn, the frequency of anomalies was determined separately among males and females. Although the rate of congenital malformations was higher in female (1.99%; 139 of 6979) than male newborns (1.68%; 120 of 7137), the difference was not statistically significant (P=0.65). The frequency of congenital anomalies categorized by gender is summarized in Table 2.

Considering the effect of congenital anomalies on the mortality rate of newborns, mortality status of unaffected newborns and affected cases were determined (Table 3). The mortality rate was significantly higher among affected cases (38.26%) than unaffected newborns (1.06%) (P<0.001). Accordingly, the prevalence of overt congenital anomalies in those born alive was 1.17% (95% CI: 1.00% to 1.35%; 163 of 13873); the rate in stillborn cases was 40.7% (95% CI: 34.61% to 46.84%; 101 of 248).

Discussion

The prevalence at birth of congenital anomalies can be defined as the number of live-born and stillborn infants with congenital anomalies, to the total number of live-births and stillbirths, in a given place and time. We used the prevalence at birth rather than the incidence of congenital anomalies to exclude affected pregnancies that end in early spontaneous abortion or pregnancy termination. Due to incomplete records in patients' charts, occult anomalies were not included in our study.

According to the current study, the central nervous system, with 139 out of 264 cases, i.e., 52.65% of all affected cases, was the most commonly affected system followed by the musculoskeletal system involving 23.86% of all congenital anomalies. Other systems with descending frequencies of congenital anomalies were the digestive tract (7.20%), the urogenital system (6.82%), chromosomal anomalies (4.92%), and the ear and neck (4.17%); we also had one case with pagus (0.38%).

Table 2. Frequency of congenital anomalies by gender (five cases not included due to ambiguous genitalia).

Gender	No. of unaffected newborns	No. of affected cases with anomalies	Total
Male	7017	120	7137
Female	6840	139	6979
Total	**13857**	**259**	**14116**

Table 3. Mortality status of all affected cases.

Mortality status	No. of unaffected newborns	No. of affected cases with anomalies	Total
Live-births	13710	163	13873
Stillbirths	147	101	248
Total	**13857**	**264**	**14121**

Comparison of reported incidences of congenital anomalies in different populations is summarized in Table 4. To be comparable to our study, we included those that included all births; that is, live- and stillbirths. In comparison with studies presented in Table 4, the frequency of congenital anomalies in our study (18.7/1000 births) was lower than that reported by Farhud et al.[8] from Tehran and Temtamy et al.[12] from Egypt; but higher than the frequencies reported in the remaining studies. Of course, it is necessary to mention that our study included only overt congenital anomalies. The different ethnic background as well as different methodologies used may be the causes of differences observed in various studies.

In the current study, although the rate of congenital anomalies was higher among females (Table 2) than males, the difference was not statistically significant (P=0.65). This finding concurs with the observation of Temtamy et al.[12] and of Lei[13] who found no correlation between the gender of the neonate and the rate of congenital anomalies either. On the other hand, it was in contrast with the findings of the study carried out in Gorgan, northern Iran, where the rate was significantly higher among male newborns.[11]

In our study, the rate of stillbirth was 1.06%

Table 4. Comparison of some reported incidences of congenital anomalies in different populations.

Population	Reference	Anomalies /1000 births
Iran (Tehran)	Farhud et al.[8]	35
Egypt (Giza)	Temtamy et al.[12]	31.7
Lebanon(Beirut)	Bittar[16]	16.5
China (west)	Cheng et al.[17]	15.4
India (Maharashtra)	Dutta and Chaturvedi[18]	12.8
Iran (Gorgan)	Golalipour et al.[11]	10.1

(147 of 13857) among the unaffected newborns and 38.26% (101 of 264) among newborns with congenital anomalies (Table 3). This significant difference (P<0.001) reflects the effect of congenital anomalies on the mortality of the newborns. Most stillbirths in the malformed cases were among those with the central nervous system defects (66.91%; 93 of 139), which is compatible with the studies carried out by Temtamy et al.[12] from Egypt, by Rasmussen et al.[14] from Atlanta, USA, and by Refaat et al.[15] in Saudi Arabia.

Considering the high frequency of central nervous system anomalies recorded in our study, it seems to be reasonable to pay more attention to the role of periconception vitamin supplementation for the primary prevention of congenital anomalies, particularly neural tube defects.

The exclusion of occult anomalies due to incomplete records was a limitation of our study, which persuade us to recommend that all neonates should be examined with scrutiny for overt as well as occult congenital anomalies. Moreover, it is necessary to establish a registry system for congenital anomalies.

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