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Congenital anomalies in Iran: a cross-sectional study on 1574 cases in the North-West of country

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Abstract

Background At least one congenital anomaly is present in between 1% and 6% of all infants throughout the world. The aim of this study was to document some epidemiological features of congenital anomalies in the North-West of Iran.

Methods The study cases ($n = 1574$) comprised all births registered/notified to three university-hospitals of Tabriz University of Medical Sciences, Iran, from 2000 to 2004.

Results Total prevalence of congenital anomalies was 165.5 per 10 000 births [95% confidence interval (CI): 157–174]. Genito-urinary tract and kidney defects, anomalies of nervous system and limb anomalies accounted proportionally for more than 65% of anomalies in the region. The total prevalence of congenital anomalies in the study area increased from 104.6 per 10 000 births in 2000 (95% CI: 90–119) to 170.1 per 10 000 births in 2004 (95% CI: 152–189).

Conclusions It is concluded that the data from this cross-sectional study of congenital anomalies in the North-West of Iran may be used as the baseline information to establish a population-based registry of birth defects in the area for health care and research purposes.

Keywords
birth defects, congenital anomalies, epidemiology, Iran

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Introduction

At least one congenital anomaly is present in between 1% and 6% of all infants throughout the world (Edmonds & James 1990; Powell-Griner & Woolbright 1990; Thein *et al.* 1992; Druschel *et al.* 1996; Johnson & Rouleau 1997; International Centre for Birth Defects 1998; Baird 1999; De Wals *et al.* 1999; Stevenson *et al.* 2000; Cordero 2002; Dastgiri *et al.* 2002; Gatt 2002; Bower *et al.* 2004a,b; EUROCAT, http://www.ihe.be/eurocat).

Because congenital anomaly is a major cause of perinatal mortality and morbidity and of disability throughout childhood and later life, this group of conditions has been subjected to intensive epidemiological scrutiny. In contrast to the large number of published research reports on the prevalence of congenital anomalies in developed countries, very few published data are available on this matter in less-developed areas.

The aim of our study was to document the epidemiological features of congenital anomalies in the North-West of Iran as the baseline information to set up a regional population-based registry of birth defects in Iran.

Methods

Congenital anomalies were defined as structural defects, chromosomal abnormalities, inborn errors of metabolism and hereditary disease diagnosed before, at, or after birth (EUROCAT working group 1997).

The main source of information was the Alzahra, Taleghani and Pediatric university-hospitals of Tabriz University of Medical Sciences, Tabriz, a major city in the North-West of Iran. In these settings, all infants are routinely examined by a gynaecologist, obstetrician, neonatologist or paediatrician at birth and hospital discharge for possible intervention and treatment. The examinations include assessment of general health, maturity and congenital anomalies. Babies born in private hospitals requiring detailed investigations, advanced medical care, surgical interventions or clinical follow-up because of a birth defect are routinely transferred to one of those university-hospitals.

The sources of ascertainment of congenital anomalies were hospital records, routine hospital discharge forms and paediatric discharge forms. The study subjects ($n = 1574$) comprised all births registered/notified to those three university-hospitals from 2000 to 2004 with a primary diagnosis under one of the following headings: nervous system anomalies, genito-urinary tract and kidney, anomalies of limb, chromosomal anomalies, cleft lip with/without palate, congenital heart disease, musculoskeletal and connective tissue anomalies, digestive system anomalies, eye and ear anomalies and other anomalies. For each infant, basic demographic information and a detailed clinical description of the major birth defect(s) were collected using the European Congenital Anomalies and Twins questionnaire (EUROCAT, http://www.ihe.be/eurocat). Pathologic confirmation of the defect was used as criteria of inclusion of fetal deaths or stillbirths in this study. We did not include malformations in terminations of pregnancy. Two midwives and a nurse were assigned in this study to code/classify the birth defects using the International Classification of Diseases (ICD)-based coding of the British Paediatric Association Classification of Disease (British Paediatric Association Classification of Disease 1979).

For inclusion in the study, an infant must have been born to a woman who resided in the defined population area of Tabriz at the time of the infant's birth. Total prevalence was then calculated by dividing the numerator (registered cases of congenital anomalies) by the relevant denominator (total live and stillbirths) for the same period of time at the same place. An infant/fetus with more than one anomaly was counted once only based on the primary diagnosis. We calculated 95% confidence interval (CI) for each prevalence rate.

Results

A total of 95 119 births were registered in Tabriz over the study period of which 93 982 (98.8%) were live births and 1137 (1.2%) stillbirths. During this period, 1574 cases with a primary diagnosis of congenital anomaly were ascertained, representing an overall prevalence rate of 165.5 per 10 000 births (95% CI: 157–174).

Table 1. Characteristics of the study subjects

	n	%
Gender		
Female	591	38
Male	910	58
Unknown	73	5
Type of delivery		
Vaginal	1059	67
Caesarean section	465	30
Other	50	3
Type of birth		
Live	1275	81
Spontaneous abortion	49	3
Induced abortion	127	8
Stillbirth	122	8
Family relation		
No relation	926	59
First cousin	329	21
Second cousin	88	6
Unknown	231	15

The basic characteristics for the subjects of the study including gender, type of delivery, type of birth and familial marriage of parents are presented in Table 1. Most of the cases were male (58%). The majority of infants were born live (81%). While there was no familial marriage in 59% of those parents with a child with congenital anomalies, first cousin marriage was observed in 21% of cases. Vaginal delivery was the main type of delivery (67%) in children with birth defects.

Table 2 shows the prevalence of birth defects in Tabriz city of Iran. Congenital anomalies with the highest rates in the region were nervous system anomalies (43.1 per 10 000 births, 95% CI: 39–47) and genito-urinary tract and kidney anomalies (39.6 per 10 000 births, 95% CI: 36–44). In contrast, the prevalence rates of musculoskeletal and connective tissue, digestive system, chromosomal, eye and ear anomalies were all less than 10 per 10 000 births.

Figure 1 shows the time trends for the prevalence (with 95% CI) of congenital anomalies over the study period. The total prevalence of congenital anomalies in the study area ranged from 104.6 per 10 000 births in 2000 (95% CI: 90–119) to 170.1 per 10 000 births in 2004 (95% CI: 152–189) with the highest rate in the birth year of 2002 (232.1 per 10 000 births, 95% CI: 211–254). The specific prevalence rate of most categories of congenital anomalies showed no consistent downward or upward trend over time from 2000 to 2004.

Table 2. Prevalence rates (per 10 000 births) of congenital anomalies by type, Tabriz (Iran)

Congenital anomaly category	Number of anomalies	Rate (per 10 000)	95% confidence interval
Nervous system anomalies	410	43.1	[39,47]
Genito-urinary tract and kidney	377	39.6	[36,44]
Anomalies of limb	251	26.4	[23,30]
Chromosomal anomalies	83	8.7	[7,11]
Cleft lip with/without palate	98	10.3	[8,12]
Congenital heart disease	129	13.6	[11,16]
Musculoskeletal and connective tissue anomalies	70	7.4	[6,9]
Digestive system anomalies	93	9.8	[8,12]
Eye and ear anomalies	30	3.2	[2,4]
Other anomalies	33	3.5	[2,4]
Total	1574	165.5	[157,174]

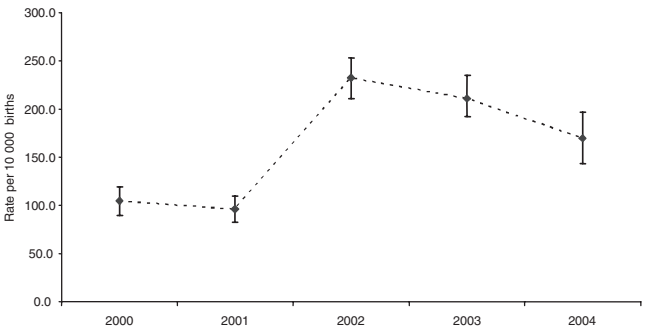

Figure 1. Prevalence of congenital anomalies with 95% CI by time trend, Tabriz (Iran), 2000–2004. CI, confidence interval.

Discussion

Despite the remarkable decline in the infant mortality rate in developed and some developing countries, birth defects account as the main factor for an increasing proportion of deaths during infancy period around the world (Powell-Griner & Woolbright 1990; EUROCAT, http://www.ihe.be/eurocat).

This investigation was a cross-sectional study designed to describe some of the epidemiological features of congenital anomalies in Tabriz, one of the major cities in Iran located in the North-West of the country. This study was conducted to provide the baseline information to set up the first regional registry of congenital anomalies in Iran. Such baseline data are essential for planning and evaluating antenatal screening and to identify clues to the aetiology of birth defects (Li *et al.* 2003).

For the birth years 2000–2004, there were a total of 95 119 births (live and still) in Tabriz, in the area covered by the Tabriz Health Board. There were 1574 cases of congenital anomalies diagnosed and notified to the three university-hospitals as having at least one of the anomalies defined/included in this study. Total prevalence of congenital anomalies was 165.5 (per 10 000 births). Genito-urinary tract and kidney defects, anomalies of nervous system and limb anomalies accounted proportionally for more than 65% of anomalies in the region.

The total prevalence of congenital anomalies in Tabriz increased from 105 (per 10 000 births) in 2000 to 170 (per 10 000 births) in 2004. However, a declining secular trend was reported for some groups of anomalies over this period. We found that the overall and specific prevalence of congenital anomalies (by different categories of defects) in Iran falls within the world range of 1–6% reported from some Middle East countries, Europe, the United States, Australia and for the world as a whole (World Health Organization 1985; Hamamy & Alwan 1994; Cordero 2002; Dastgiri *et al.* 2002; Bower *et al.* 2004b; EUROCAT, http://www.ihe.be/eurocat).

It should be mentioned that the rates reported in this study might have underestimated the prevalence of birth defects in the region as not all congenital anomalies could be detected at birth or shortly thereafter because of the lack of cytogenetic and teratology investigations or autopsies for stillbirths and neonatal deaths. We could not also include anomalies in terminations of pregnancy. Some defects may not even be diagnosed in the first year after birth as they are less severe and clinically undetectable. However, for validation purposes, the study subjects were assessed/diagnosed by the specialized and trained clinical staff including obstetrician, gynaecologist, neonatologist or paediatrician. And there was also a reliable recording and monitoring system of major and clinically identifiable anomalies of public health importance at birth or soon after in the study population.

In seeking to explain the upward pattern of prevalence of congenital anomalies in the birth year of 2002, potential underlying contributing factors should be considered. These factors include case ascertainment methods, data collection and

sources of information which might have varied over time and/or in those three hospitals. In addition, although unproven, the possible impact of some specific local risk or causal factors on this pattern including changes in environmental, nutritional and lifestyle variables (Kalter & Warkany 1983b; Eizaguirre-Garcia *et al*. 2000; Kalter 2000; MacDonnell *et al*. 2000) can not be ruled out. A supportive evidence for this explanation is a similar time trend for Guillain–Barre syndrome which has recently been reported in the same population of our study (Departments of Epidemiology, Paediatrics and Neurology. Tabriz University of Medical Sciences, Tabriz, Iran, Unpublished Data from an Ongoing Research Project: 2001–2004).

Despite some limitations, our study provides basic information on the magnitude and spectrum of public health problems caused by different types of congenital anomalies identified and diagnosed at birth or soon afterwards in the North-West of Iran. These data should also be essential to establish a national registration scheme for congenital anomalies and genetic disorders.

Our study should hopefully provide an opportunity for healthcare authorities in the area and a necessity to develop innovative health education strategies, standard screening and diagnostic procedures during the perinatal period. Moreover, it is imperative to develop healthcare facilities and effective support educational, social and rehabilitation services for those children surviving with a disability, impairment or handicap as a result of birth defects.

Finally, as it is estimated that up to 60% of congenital anomalies are preventable (Czeizel *et al*. 1993), larger longitudinal epidemiological studies and randomized clinical trials are required at the community level to evaluate the impact of preconceptional and prenatal diet supplementation with multivitamins and other micronutrients as preventive measures of congenital anomalies (Czeizel 1993) that are currently being undertaken by the Ministry of Health in some parts of the country.

In conclusion, the data from this cross-sectional study of congenital anomalies in the North-West of Iran may be used as the baseline information to establish a population-based registry of birth defects in the area for health care and research purposes.

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