brief report

Profile of major congenital anomalies in the Dhahira region, Oman

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About 2 to 3% of births are associated with major congenital anomalies diagnosed at or soon after birth. Congenital malformations accounted for an estimated 495 000 deaths worldwide in 1997. The great majority of these deaths occurred during the first year of life, and thus tend to contribute mostly to the infant mortality rate. Because of the multiple factors involved, infant mortality attributable to congenital anomalies can be expected to vary substantially among countries.[1] Children with congenital anomalies who survive the high-risk period during the first year of life still have low survival rates as children and adults. The leading causes of infant morbidity and mortality in poorer countries are malnutrition and infections,[2] whereas in developed countries they are accidents and congenital malformations.[3] With the control of infectious diseases and malnutrition, particularly in developed countries, congenital anomalies are now making a proportionally greater contribution to ill health in childhood.[4]

Worldwide surveys have shown that the frequency of birth defects varies greatly from country to country. The frequency depends on the time of observation after birth, the types of malformation included, and the differences in reporting and statistical procedures.[5] National vital statistics data from the USA indicates that congenital anomalies significantly contribute to the mortality of children, and nearly 11% and 6% of deaths in children ages 1-4 years and 5-9 years, respectively, were attributable to congenital anomalies in 1999.[6] Despite declining prevalence, they are also still a major cause of perinatal mortality and childhood disability in Europe.[7] Congenital anomalies are an important cause of fetal, neonatal, and child mortality and morbidity, accounting for 21% of perinatal and infant deaths in the United Kingdom in 2001.[8] In the Dhahira region, an effective and comprehensive vaccination programme is in place, childhood malnutrition has declined and serious childhood infections are disappearing. Consequently, congenital anomalies began to emerge as one of the major childhood health problems.

Treatment and rehabilitation of children with congenital anomalies is costly, and complete recovery is usually not possible.[9] Hence, it is an obligation to find out the causative and risk factors for congenital anomalies and prevent them at the earliest. The causes of the majority of congenital anomalies are not currently understood. A combination of genetic, biologic or environmental factors is considered to be responsible for many of these conditions. However, the cause is not known (cause not ascertainable) in about 40% to 60% of cases in general.[10]

Al Dhahira region is in the northwest of Oman. It has a population of 207 015 distributed in five Wilayats (districts) and approximately 51% of the female population is of child-bearing age (15-45 years) (census 2003). During 2003 to 2005, there were 10 311 births in the Dhahira region Ministry of Health institutions (MOH), resulting in 10213 live births and 98 still births.[11] Congenital anomalies accounted for 20.5% of fetal deaths in Oman during the year 2004.[12] Of the 21 988 births during the study period, 541 babies (24.6 per 1000 births) had major congenital malformations in a study conducted in Nizwa, Oman.[13]

Prevalence studies of congenital anomalies are useful to establish baseline rates, to document changes over time, and to identify clues to etiology. They are also important for health services planning and evaluating antenatal screening for congenital anomalies, particularly in high-risk populations.[7] There may be regional variations in the pattern of congenital anomalies and a similar sort of study has not been conducted in Dhahira province in the north of the country. The present study was conducted to provide prevalence and a descriptive overview of the congenital anomalies in the Dhahira region.

METHODS

There are 10 health institutions with a maternity facility including two secondary care hospitals in the Dhahira region. Almost all the deliveries take place in these institutions and seek health care there after. This cross-sectional retrospective record-based study was conducted from January 2003 to December 2005 in the region. Congenital anomaly registration covers approximately 3500 births every year in the Dhahira region. Notification of anomalies in live, stillbirths and induced abortions to the regional office is mandatory and usually done through a standard notification form (H/P-4) filled in by health professionals across the MOH institutions in the Dhahira region, which was started officially in the year 2003.

Each notification form collects information on all congenital anomalies occurring in miscarriages after 20 weeks of gestation, in live births and stillbirths, and in fetuses that are terminated after prenatal diagnosis of a life-threatening anomaly. All the major defects for which the degree of ascertainment is high and classified specific codes were included in the study. Variables recorded include child and family information (Section A) (name, date of birth, demographic data, nationality, consanguinity and maternal age), medical information (Section B) (birth status, plurality, sex, weight, gravida and diagnostic methods) and diagnosis (Section C) (ICD-10 codes, anomalies, registration details). No autopsy examinations were performed on those newborns that died after birth.

Congenital anomalies were defined as structural defects, chromosomal abnormalities, inborn errors of metabolism, and hereditary disease diagnosed before, at, or after birth. The diagnostic methods used were clinical, radiography, CT scan, ultrasound, laboratory tests and cytogenesis wherever necessary in ascertainment of cases.

Cases comprised congenital anomalies identified in live births (LB), stillbirths (SB), and induced abortions (IA) following prenatal diagnosis. Total prevalence rate was calculated by the number of cases among LB+SB+IA / by number of total births x 1000. An infant/fetus with more than one anomaly was counted once only based on the primary diagnosis for calculating the overall prevalence rate.[14] Birth weight <2500 g was considered low birth weight (LBW).

The data was computed and were analyzed using the Statistical Package for Social Sciences (SPSS, version 9). Rates and proportions were calculated with 95% confidence intervals. The proportions were compared using the test of proportions and the chi-square test. The level of significance was $P<0.05$.

RESULTS

Over the 3-year study period, 10 311 newborns were delivered in the region, including 10,213 (99%) were live births and 98 (1%) stillbirths. During this period, 84 babies with a primary diagnosis of congenital anomaly were ascertained, with an overall prevalence rate of 8.1 per 1000 total births (95% CI, 6.6-10.1). Of the 84 babies, 81 (96.4%) were live births and 3 (3.6%) stillbirths. The majority of the mothers who gave birth to babies with a congenital anomaly were Omani women (96.4%) and belonged to Ibri Wilayat (45.2%).

The mean gestational age at delivery was 37.1±3.5 weeks and 32.2% of the mothers had a gestational age < 37 weeks (preterm). Nearly 79% of the mothers were multigravida (>2 gravida) and 29.8% of the mothers were grand multiparous (parity 8 or more). The total mean weight was 2879.3±655.9 g with 19% of the fetuses weighing < 2500 g (LBW) as shown in Table 1.

About 63% (53) of the delivered fetuses were males and 3.5% (3) were ambiguous. The rate of congenital

Table 1. Distribution of congenital anomalies according to maternal and fetal characteristics.

Maternal characteristics (N=84)	Number	Percent	Mean±SD
Age (years)			
< 25	21	25.0	30.0±7.3
25-30	21	25.0	
30-35	16	19.0	
≥ 35	26	31.0	
Gestational age (weeks)			
20-27	2	2.4	37.1± 3.5
28-36	25	29.8	
≥ 37	57	67.8	
Parity			
0	2	2.4	5.2± 4.0
1-4	39	46.4	
4-8	18	21.4	
≥ 8	25	29.8	
Birth weight (g)			
500-2499	16	19.1	2879.3±655.9
2500-2999	29	34.5	
3000-3499	29	34.5	
≥ 3500	10	11.9	

Table 2 Congenital anomalies (CA) among the study subjects by gender and nationality.

	No. of babies without CA	No. of babies with CA	Percent	Relative risk	95% CI
Gender *					
Male	5303	53	1.0	1.78	1.13 – 2.82
Female	5005	28	0.56	reference	-
Nationality					
Omani	9611	81	0.84	0.50	0.16 – 1.60
Non-Omani	700	3	0.43	reference	-

* 3 babies had ambiguous genitalia

anomalies was significantly different between male and female babies ($P<0.05$). The prevalence of congenital anomalies was twice as high among Omani mothers (8.5/1000 births) when compared with non-Omanis (4.3/1000 births), but the difference was not statistically significant ($P>0.05$) (Table 2). Three of 84 (3.5%) patients with a congenital anomaly died, although the total deaths among the 10 311 babies born were 95 (0.92%). The death rate was relatively higher in patients with congenital anomalies ($\chi 2$ [Yates corrected] =3.69, $P=0.05$, odds ratio=3.95).

The common congenital anomalies identified in the Dhahira region are shown in Table 3. Eighty-four babies had 117 congenital anomalies. Approximately 24% (20/84) of babies had multiple anomalies. The leading congenital anomalies were musculoskeletal (23%, 27/117) followed by anomalies of the circulatory (17.1%) and genitourinary (15.4%) systems. The single most common anomaly was hypospidiasis (8.5%, 10/117) followed by Down's syndrome (7.7%). Chromosomal abnormalities accounted for 10.3% (12/117) of the anomalies.

The consanguinity rate was high among parents of babies with congenital anomalies (73.8%), with 54.1% married to first cousins.

DISCUSSION

The total prevalence of congenital anomalies in this study was low compared with other studies conducted in this part of the world.[13,15] Table 4 illustrates the prevalence rates in different parts of the world compared with our study. The *World Atlas of Birth Defects* published by the World Health Organization (WHO) gives an overview of prevalence rates around the globe.[20] The wide variation in the congenital anomaly rates could be due to different case ascertainment methods (including data collection, sources of information, and type of notification, inclusion and exclusion criteria), demographic and environmental factors and definitions used in a particular region.

Some of the variation between anomalies is explained by the fact that those obvious at birth (such as cleft lip, limb defects) are more likely to be ascertained than "hidden" defects (such as renal anomalies, cardiac defects), which may be diagnosed later. Under-ascertainment in congenital anomalies has been a long-known problem.[21] In this study, the fetal death among anomaly babies was 3.5%, in contrast to the national fetal death report, which revealed that congenital anomalies were present in 20.5% of the fetal deaths in Oman,[12] which reflects that some of the fetal deaths with congenital anomalies as a risk factor are being reported as only fetal deaths but not as congenital anomalies. Thus, there is a need to strengthen the surveillance of congenital anomalies and report fetal death with congenital anomalies in both the fetal death and congenital anomaly forms separately. Consequently, the prevalence of congenital anomalies in our study is an underestimate in the Dhahira region.

The causes of congenital anomalies are often complex and at times the cause is unknown. Maternal and environmental factors are strongly associated with congenital anomalies. Among the various possible risk factors studied, a higher incidence of congenital malformations were associated with increasing maternal age (>35 years), higher gravida mothers (>G4), parental consanguineous marriages, a previous history of abortions, and maternal hypertension.[22] Research has shown that advanced maternal age (≥35 years) is a risk factor for chromosomal abnormalities, especially Down's syndrome.[23] In our study, women with a maternal age ≥35 years (31.0%) and gravida ≥4 (51.2%) years had a higher proportion of congenital anomalies when compared to other groups (Table 1). Risk factors such as maternal smoking, hypertension, a previous history of abortion, obesity and chemical exposure need to be included in the congenital anomaly notification forms

Table 3. Prevalence rate (per 1000 births) of various congenital anomalies in the Dhahira region.

Anomalies documented	Number	Percent	Prevalence rate	95 % CI
Neurological	10	8.5	1.0	0.5 – 1.8
Anencephaly	1	0.9		
Microcephaly	6	5.1		
Congenital hydrocephalus	3	2.6		
Eye, ear and neck	7	6.0	0.7	0.3 – 1.4
Circulatory	20	17.1	1.9	1.3 – 3.0
Malformations of cardiac chambers and connections	6	5.1		
Cardiac valve	2	1.7		
Great vessels	5	4.3		
Septal defects	7	6.0		
Cleft lip and palate	14	12.0	1.4	0.8 – 2.3
Cleft palate	6	5.1		
Cleft lip	3	2.6		
Cleft palate and lip	5	4.3		
Digestive system	5	4.3	0.5	0.2 – 1.1
Genital and urinary	18	15.4	1.7	1.1 – 2.8
Undescended testes	3	2.6		
Hypospidiasis	10	8.5		
Ambiguous genitalia	3	2.6		
Others	2	1.7		
Musculoskeletal	27	23.1	2.6	1.8 – 3.8
Hip deformities	2	1.7		
Feet deformities	5	4.3		
Head, spine and chest deformities	5	4.3		
Polydactyly	5	4.3		
Syndactyly	3	2.6		
Skeletal displasias	3	2.6		
Others	4	3.4		
Multiple system syndromes	4	3.4	0.4	0.2 – 1.0
Chromosomal	12	10.3	1.2	0.7 – 2.0
Down's syndrome	9	7.7		
Edwards's and Patau's syndrome	3	2.6		
Total	117		11.3	9.5 – 13.6

Table 4 Comparison of congenital anomaly rates in various countries.

Congenital anomaly prevalence rate/1000 births	Study
9.2	Present study
10.5	Reference 15
12.9	Reference 16
15.4	Reference 17
17.0 (incidence)	Reference 18
24.6	Reference 13
32.0	Reference 19

(H/P-4). Risk factor studies and additional research into the pathophysiology is needed to understand the causes and prevent congenital anomalies.

The rate of anomalies in male babies is nearly twice that of females, which might be explained, in part, by the fact that X-linked disorders will be only apparent in males who will show the phenotype. This result is the same as reports from Iran.[24-25] As in studies from Iran, India and Singapore, the musculoskeletal system was the most frequently affected system in our study, followed by cardiovascular system defects and urogenital system and central nervous system defects.[25-27] However, other studies have reported gastrointestinal,[15] central nervous system and cardiovascular system defects, as the most frequently affected systems.[28-29]

A higher prevalence of 31% and 19.6% of chromosomal abnormalities has been reported from studies conducted elsewhere compared with our study (10.3%, 12/117).[15,27] Likewise, Down's syndrome was less common (1 in 1135 live births) compared to a study conducted by (1 in 700 live births) Thein et al.[27] Neural tube defects are one of the leading causes of fetal and infant mortality caused by congenital anomalies around the world. Unlike other studies,[30] neural tube defects were low in our study.

Finally, to support the genetic causes of congenital anomalies, consanguinous marriages in our study were 73.8% and of those, 54.1% had married first cousins, which is higher than the national average.[31] Studies by Naderi[32] and Khrouf et al[33] have also reported a higher rate of congenital anomalies among consanguineous parents. Pre- and postmarital genetic counselling is considered necessary since the consanguinity is high among Arab Muslims in this part of the world.[34]

This descriptive epidemiological study of congenital anomalies in the Dhahira region indicates a reasonably low prevalence, which is suggestive of underascertainment of congenital anomalies in the region. Efforts to enhance the surveillance and further reduce the congenital anomalies should be directed at prevention, especially premarital and preconceptional counselling.

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