The National Congenital Anomalies Register in the United Arab Emirates

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السجل الوطني للشذوذيَّات الخِلقية في دولة الإمارات العربية المتحدة

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الخلاصة: هذا السجل الذي بدأ العمل به في شهر كانون الثاني/يناير 1999، هو سجل سُكَّانيٌّ الـمُرْتَكَز، يغطي كافة حالات الولادة في دولة الإمارات العربية المتحدة. وقد كان الغرض من إجراء هذه الدراسة، تقييم التقدُّم الحاصل في أعمال هذا السجل، وتحديد مدى انتشار الشذوذيَّات الخِلقية وعوامل الاختطار المرتبطة بذلك على كلٍّ من الأم والوليد. وكان المعدل الإجمالي لانتشار الشذوذيَّات الخلقية خلال الفترة من 1999 إلى 2001 هو: 7.89، و10.95 و7.92 لكل ألف مولود حي، على التوالي، بين المولودين أحياء والمولودين مَوْتَى ومجموع المواليد. وهي معدلات قابلة للمقارنة مع المعدلات العالمية لجميع مناطق دولة الإمارات، باستثناء دبي والفجيرة ورأس الخيمة. ووفقاً للتصنيف الدولي للأمراض، فإن الجهاز القلبي الوعائي كان أكثر الأجهزة تأثُّراً، تلته الشذوذيَّات الخلقية الكروموسومية المنشأ وشذوذيَّات الجهاز الهيكلي العضلي. وكانت العيوب الولادية أكثر شيوعاً لدى الأمهات الأكبر عمراً، وفي الأمهات المتكررات الولادة، وبين الذكور، وناقصي الوزن، والخدَّج.

ABSTRACT The National Congenital Anomalies Register is a population-based register covering all births in the United Arab Emirates. We evaluated the progress of the register and determined the prevalence of congenital anomalies (CAs) and associated maternal and neonatal risk factors. Total prevalence of CAs for 1999–2001 was 7.89/1000, 10.95/1000 and 7.92/1000 for live births, stillbirths and total births respectively. Rates were comparable to international rates for all districts except Dubai, Fujairah and Ras Al Khaimah. According to the ***International classification of diseases***, the cardiovascular system was the most affected followed by CAs of chromosomal origin and the musculo-skeletal system. Birth defects were more common with older maternal age, grand multiparity, male babies, low-birth-weight babies and premature babies.

Le Registre national des anomalies congénitales aux Émirats arabes unis

RÉSUMÉ Le Registre national des anomalies congénitales est un registre dans la population qui couvre toutes les naissances survenant aux Émirats arabes unis. Nous avons évalué l'état d'avancement du registre et déterminé la prévalence des anomalies congénitales et des facteurs de risque associés pour la mère et le nouveau-né. La prévalence totale des anomalies congénitales pour la période 1999-2001 était de 7,89 pour 1000, de 10,95 pour 1000 et de 7,92 pour 1000 pour les naissances vivantes, les mortinaissances et les naissances totales respectivement. Les taux étaient comparables aux taux internationaux pour tous les districts sauf pour Dubai, Fujairah et Ras Al Khaimah. Suivant la ***Classification internationale des maladies***, le système cardio-vasculaire était le plus touché, suivi par les anomalies congénitales d'origine chromosomique et du système ostéo-articulaire et des muscles. Les malformations congénitales étaient plus fréquentes lorsque la mère avait un âge plus avancé, en cas de grande multiparité, chez les garçons, les bébés de faible poids de naissance et les prématurés.

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Introduction

The impact of major birth defects on the fetus and newborn infant is great: this is now one of the leading causes of perinatal death, both in industrialized and developing countries [*1*].

The infant mortality rate was 8.21 per 1000 live births in the United Arab Emirates in the year 2000 [*2*]; of 451 infant deaths, 339 (75.3%) were due to congenital anomalies (CAs). This rate is one of the highest in the world and can be explained by the unique demographic and medical health care situation in the United Arab Emirates [*1,3*].

According to the World Health Organization, the term congenital anomaly includes any morphological, functional and biochemical-molecular defects that may develop in the embryo and fetus from conception until birth, present at birth, whether detected at that time or not, and this term is synonymous with the term birth defect used in the United States of America [*4,5*].

Congenital anomalies represent a special category of disorders characterized by early onset and limited chance of complete recovery. Prevention, which is the optimal solution, is based on an understanding of the causes and possible risk factors of CAs. Appropriate datasets, including baseline prevalence of different types of CA, are therefore essential prerequisites for prevention programmes [*6,7*]. Hence, the Ministry of Health, through the Department of Maternal and Child Health, set up the National Congenital Anomalies Register in the United Arab Emirates.

In the preparatory phase of the register, 2 studies were performed to obtain baseline birth prevalence of CAs. The first was a pilot study done over 3 months in 1998, the purpose of which was to try out and finalize the notification form in 3 districts (Abu-Dhabi, Al-Ain and Western Region) [*8*]. The second was a hospital-based study in Al-Mafraq Hospital, a major hospital in Abu-Dhabi, evaluating the data collected in 1992–1994 to check the quality and to calculate the total baseline prevalence of CA and prevalence of different types and compare the rates with international figures [*9*].

The National Congenital Anomalies Register is a population-based register covering all births in the United Arab Emirates and was launched in all medical districts in January 1999. The expected benefits of the register are to determine the baseline birth prevalence of each type of CA, to increase awareness within the medical community about these conditions, and to improve the quality of diagnosis and recording. The aim is to establish a priority list of preventive measures and help to organize better care for patients with special needs, e.g. those with Down syndrome. In addition, the register has a surveillance function because the identification of significant changes in the baseline rate for specific CAs may be an indication of peculiar personal or demographic characteristics, e.g. a high proportion of women of advanced age or a high rate of consanguinity.

The aim of this study was to evaluate the progress of the register and to determine the prevalence of different types of CAs and the associated maternal and neonatal risk factors.

Methods

The National Congenital Anomalies Register is a system of ongoing, permanent registration for the collection, storage and analysis of personal, demographic and medical data on affected neonates and infants. For the purposes of the register, CAs are defined as structural defects of fetal

development that require medical treatment. Minor anomalies such as hydrocele, coronal hypospadia and preauricular tag are therefore excluded [*10*].

Coding of CAs is defined as in chapter XIV of the 9th version of the *International classification of diseases*, codes 740.0–759.9, with slight modification in multiple CAs. The anomalies were classified as isolated or multiple because the unit of evaluation was the person affected, i.e. index cases [*10,11*].

The purpose of the register is to record all liveborn infants (from birth to 1 year) and stillborn fetuses having CAs. Abortion is not legal in the United Arab Emirates, therefore selective termination of malformed fetuses is not permitted. The main reporting sites of CAs are obstetric units (98% of deliveries take place in maternity hospitals), paediatric clinics and departments, and maternal and child health centres.

A special notification form is used to collect required data. Reported data, mainly the diagnosis of CA, is critically evaluated and coded at the Department of Maternal and Child Health in the Ministry of Health.

The data were used to determine the prevalence of CAs at the national level for live births and stillbirths during the period 1999–2001. Rates were also calculated for each medical district. The results were compared with other studies done in the United Arab Emirates and in some countries of the Eastern Mediterranean Region. To determine the associated risk factors and their relationship to CAs, we studied the rate and degree of consanguinity, some maternal factors (age and parity) and some infant factors (sex, birth weight and gestational age). Statistical analysis was done using *SPSS*, version 8.

Results

A total of 164 244 births have been recorded since the implementation of the register (January 1999–December 2001).

Table 1 shows the prevalence of CA according to pregnancy outcome for both United Arab Emirates nationals and non-nationals. The overall prevalence for CA during the study period was 7.9 per 1000 total births, 8.0 per 1000 live births and 11.0 per 1000 stillbirths. The yearly prevalence of birth defects for live births and total births was almost the same over the 3 years of the study. The yearly prevalence for stillbirths, however, increased from 5.4/1000 in 1999 to 14.4 in 2001.

Table 1 **Prevalence of reported congenital anomaly (CA) according to pregnancy outcome, 1999–2001**

Year	Pregnancy outcome								
	Live births			Stillbirths			Total births		
	No.	No. CA	No./ 1000	No.	No. CA	No./ 1000	No.	No. CA	No./ 1000
1999	51 580	415	8.1	370	2	5.4	51 950	417	8.0
2000	53 807	384	7.1	424	5	11.8	54 231	389	7.2
2001	57 578	487	8.5	485	7	14.4	58 063	494	8.5
Total	162 965	1286	8.0	1279	14	11.0	164 244	1300	7.9

Table 2 **Prevalence of reported congenital anomaly (CA) for each medical district, 1999–2001**

Medical district	Total births	CAs, No.	No./ 1000
Abu Dhabi	40 489	431	10.6
Al Ain	26 425	270	10.2
Western Region	3 655	51	14.0
Dubai	48 599	31	0.6
Sharjah	21 166	358	16.9
Ajman	5 816	63	10.8
Umm Al Quwain	2 146	31	14.5
Ras Al Khaimah	9 315	41	4.4
Fujairah	6 633	24	3.6
Total	164 244	1300	7.9

Table 2 gives the prevalence of CA for each district of the United Arab Emirates, for both nationals and non- nationals, during the study period. There was an appreciable difference in the rate for reported cases of CA between the 9 medical districts. The highest prevalence was reported in Sharjah (16.9/1000 total births) followed by Umm Al Quwain (14.5/1000 total births) while the lowest was in Dubai (0.6/1000 total births) followed by Fujairah (3.6/1000 total births).

Table 3 shows the prevalence for different categories of CA for United Arab Emirates nationals according to pregnancy outcome during the period 1999–2001. The overall rate was 6.5/1000 total births (6.4/1000 live births and 18.5/1000 stillbirths). Of the 441 reported cases of CA, 296 were isolated (67.1%) and 145 multiple (32.9%). The cardiovascular system was the most affected (1.2/1000 total births), followed by CAs of chromosomal origin and of the musculoskeletal system (0.8/1000 total births for each). The prevalence of neural tube defect was 0.5/1000 total births. The CA incidence in any subgroup did not differ for live births or total births (Table 3).

The most commonly reported CA for stillbirths was neural tube defect (5.6/1000 stillbirths). It was noted that 16 out of 34 reports of neural tube defect were from Abu-Dhabi medical district, prevalence 0.9/1000 total births.

The consanguinity rate for United Arab Emirates nationals with CAs was 52.7%.

Table 4 shows some maternal and infant risk factors for nationals and their relationship to CAs. The prevalence of CA was 5.0/1000 total births for mothers who had given birth to 2 or fewer children and increased to 7.3/1000 total births for those had given gave birth to 5 or more children ($P = 0.007$). The incidence for CA increased from 4.5/1000 total births for mothers 16–19 years to 7.4/1000 total births for mothers 40 years or older. The association between maternal age and CA prevalence, however, is not statistically significant ($P = 1.000$).

There seems to be a positive correlation between chromosomal CAs and both parity and maternal age. In babies born to mothers 40 years or older, 25.5/1000 total births had chromosomal CAs while only 5.2/1000 total births had non-chromosomal CAs. Similarly, in babies born to mothers with higher parity (parity ≥ 5) 49.1/1000 total births had chromosomal CAs compared to 33.4/1000 total births who had non-chromosomal CAs.

Prevalence of CA was 6.9/1000 total births for males and 5.2/1000 total births for females. The rate for neonates with birth weight ≥ 2500 g was 4.5/1000 total births while the rate for those with birth weight < 2500 g (low birth weight) was 19.2/1000 total births. The data on birth weight variables include only the year

Table 3 **Prevalence of different categories of congenital anomaly (CA) according to pregnancy outcome for United Arab Emirates nationals, 1999–2001**

Type of CA	ICD code	Pregnancy outcome					
		Live births		Stillbirths		Total births	
		CAs, No.	No./ 1000	CAs, No.	No./ 1000	CAs, No.	No./ 1000
Isolated CAs		290	4.3	6	11.1	296	4.3
Neural-tube defects	740.0–742.0	31	0.5	3	5.6	34	0.5
Other CAs of nervous system	742.1–742.9	16	0.2	1	1.9	17	0.2
CAs of eye	743.0–743.9	3	0.0	0	0.0	3	0.0
CAs of ear, face and neck	744.0–744.9	5	0.1	0	0.0	5	0.1
CAs of cardiovascular system	745.0–747.9	84	1.2	1	1.9	85	1.2
CAs of respiratory system	748.0–748.9	2	0.0	0	0.0	2	0.0
Orofacial cleft	749.0–749.3	19	0.3	1	0.0	20	0.3
CAs of digestive system	750.0–751.9	14	0.2	0	0.0	14	0.2
CAs of genital organs	752.0–752.9	40	0.6	0	0.0	40	0.6
CAs of urinary system	753.0–753.9	20	0.3	0	0.0	20	0.3
Certain CAs of musculoskeletal system	754.0–754.8	21	0.3	0	0.0	21	0.3
Other CAs of limbs	755.0–755.9	19	0.3	0	0.0	19	0.3
Other CAs of musculoskeletal system	756.0–756.9	14	0.2	0	0.0	14	0.2
CAs of integument	757.0–757.9	2	0.0	0	0.0	2	0.0
Multiple CAs		141	2.1	4	7.4	145	2.1
Gene CAs	756.4, 759.4, 759.5	29	0.4	0	0.0	29	0.4
Chromosomal CAs	758.0–758.9	55	0.8	0	0.0	55	0.8
Other multiple CAs	759.2, 759.3, 759.8	3	0.0	1	0.0	4	0.1
Unspecified multiple CAs	759.7	54	0.8	3	5.6	57	0.8
Total	–	431	6.4	10	18.5	441	6.5

ICD = International classification of diseases.

1999. Prevalence of CA for full term babies was 4.7/1000 total births while for premature babies it was 28.9/1000 total births.

Discussion

Congenital anomaly occurs in 1 out of every 28 births, affecting millions of families. These defects are the leading cause of infant death; they also have a profound effect on the daily lives of people of all ages [*12*].

Our data indicate that the prevalence of CAs for both nationals and non-nationals during the period 1999 to 2001 was 8/1000 live births and 7.9/1000 total births (Table 1). The figures for United Arab Emirates nationals during the same period were 6.4/1000 live births, and 6.5/1000 total births (Table 3). These figures are comparable to, or somewhat lower than, results of studies from the Libyan Arab Jamahiriya with 7.0/1000 live births and 9.3/1000 total births [*13*], Bahrain with 7.2/1000 total births in 1978 and 18.5/1000 total births in 1985 [*14*] and Saudi Arabia with 8.5/1000 live births [*15*].

Table 4 **Classification of congenital anomaly (CA) cases of total births (live births + stillbirths) for United Arab Emirates nationals according to some maternal and some infant variables during the period 1999–2001**

Variable	Total births	CAs, No.	No./1000	%[a]	χ^2 (*P*-value)
Parity					
1–2	22 948	115	5.0	26.1	9.993 (0.007)
3–4	18 256	103	5.6	23.4	
≥ 5	21 460	156	7.3	35.4	
Not stated	5 485	67	12.2	15.2	
Total	68 149	441	6.5	100.0	
Maternal age (years)					
16–19	4 676	21	4.5	4.8	0.406 (1.000)
20–24	13 036	92	7.1	20.9	
25–29	18 028	90	5.0	20.4	
30–39	22 812	124	5.4	28.1	
40+	4 608	34	7.4	7.7	
Not stated	4 989	80	16.0	18.1	
Total	68 149	441	6.5	100.0	
Sex of infant[b]					
Female	32 904	172	5.2	39.0	7.570[c] (0.006)
Male	35 245	242	6.9	54.9	
Undefined	–	1	–	0.2	
Not stated	–	26	–	5.9	
Total	68 149	441	6.5	100.0	
Birth weight[d]					
Low birth weight	1 771	34	19.2	27.6	62.020 (< 0.001)
Birth weight > 2500 g	19 807	89	4.5	72.4	
Total	21 578	123	5.7	100.0	
Gestational age					
Premature	3 596	104	28.9	23.6	323.300 (< 0.001)
Full term	60 318	282	4.7	63.9	
Not stated	4 235	55	13	12.5	
Total	68 149	441	6.5	100.0	

[a]As % of all congenital anomalies.
[b]Undefined means an infant of indeterminate sex; not stated is due to error in the registration of the congenital anomalies forms. Only male and female births are registered in the annual statistical report issued by the Ministry of Health.
[c]Chi-squared was computed after excluding not stated and undefined.
[d]Year 1999 only.

Our figures are also in accordance with reported figures in many industrialized and developing countries [*16*]. The figures in the stillbirth group (11.0/1000 nationals plus non-nationals, 18.5/1000 nationals) are much lower than global figures, the latter reaching 100–200/1000 stillbirths [*6*].

Our results might, however, be an underestimate which could be attributed to incomplete cooperation between the national

register and some emirates, lack of post mortem examinations for stillbirths and neonatal deaths and underestimation of cases with malformation, especially certain internal malformations.

There were clear differences in the prevalence of reported CAs among the 9 medical districts (Table 2). It was comparable to international figures for all medical districts except for Dubai, Fujairah and Ras Al Khaimah. This can be explained by the underreporting of cases in these districts. About 30% of total births took place in Dubai, and by excluding Dubai, the prevalence of reported CAs would be 9.8 per 1000 total births and 9.7 per 1000 live births for United Arab Emirates nationals. The underreporting of malformed cases by some districts can be explained by certain administrative and communication barriers with those districts.

Investigations of CAs in early pregnancy are limited to ultrasound and maternal serum α-fetoprotein if required. Ultrasound examination is the only investigation done routinely for pregnant women, and maternal serum α-fetoprotein is done only if there is a family history of neural tube defect. Maternal serum multiple-marker screening (including fetoprotein) and more detailed ultrasound examination for all pregnant women would increase the number of reported CAs for both live births and stillbirths.

The prevalence of CA for citizens in Abu Dhabi medical district (10.0/1000 total births) and Al-Ain medical district (11.7/1000 total births) are comparable to the figures reported in earlier studies done in Abu Dhabi medical district with an incidence of 12.9/1000 total births [*17*] and Al-Ain medical district with an incidence 10.5/1000 total births [*18*].

Of the 441 reported cases with CA in United Arab Emirates nationals, 296 (67.1%) were isolated and 145 (32.9%) were multiple CAs (Table 3). The proportion of multiple anomalies was higher than the international figure, which is 15% of total anomalies [*10*]. Further research needs to be done in this field because of the high rate of consanguinity [*19*].

Using the *International classification of diseases* classification, the cardiovascular system was the most affected (1.2/1000 total births), followed by CAs of chromosomal origin and the musculoskeletal system (0.8/1000 total births for each). The prevalence of neural tube defects was 0.5/1000 total births (Table 3). Our results were in line with observations in Saudi Arabia where the cardiovascular system was the most commonly affected system followed by the musculoskeletal system, gastrointestinal system, chromosomal causes and neural tube defects [*15*].

Although cardiovascular CAs were the largest proportion among isolated CAs, the distribution of isolated CAs did not indicate any particular cluster. We found that 12.9% of CAs were categorized as unspecified multiple CAs, indicating limitations of accurate genetic diagnosis in some cases.

Ten out of 441 CA cases were stillbirths, only 3 of them diagnosed as neural tube defects (Table 3). This was owing to the low inclusion of stillborn infants in the national register, failure to conduct post mortem examinations and the increase in awareness about using folic acid during pregnancy. The prevalence of neural tube defects in Abu Dhabi medical district for United Arab Emirates citizens was 0.9/1000 total births. This figure is comparable to, or lower than, other studies done in Abu Dhabi medical district with an incidence ranging from 0.99/1000 total births [*17*] to 1.13/1000 total births (G.R. Samson, unpublished data, 2001) and 1.77/1000 total births (M.R. Sedaghatian, unpublished

data, 2001). The high number of neural tube defects reported from Abu-Dhabi medical district can be explained by good reporting quality for all CAs, including neural tube defects, owing to better investigative facilities.

Concerning the associated risk factors and their relationship to the CAs, our study showed that the consanguinity rate for citizens with CAs was 52.7%, which is not different from the consanguinity rate (51%) in the population as a whole [*19*].

The incidence for births with CAs was increased for mothers who had had 5 or more children (1.5 times the rate for mothers with 2 or fewer children) and mothers of 40 years or older (1.5 times the rate for mothers under 20 years) (Table 4). Older maternal age and grand multiparity have been implicated elsewhere with higher occurrences of birth defects [*20*].

We found the proportion of infants who had chromosomal compared to non-chromosomal CA was higher among those born to mothers of 40 years or older and of high parity (5 or more). This is comparable to the findings of a study done in Saudi Arabia where the proportion of infants born with chromosomal CA was 6 times higher than those with non-chromosomal CA for mother ≥ 40 years and 2 times higher for those born to mothers of higher parity (parity ≥ 5)[*15*].

Birth defects were more common among males (6.9/1000 births) than females (5.2/1000 births). Low-birth-weight babies (< 2500 g) were about 4 times more likely to have CAs than babies of normal birth weight and premature babies were about 6 times more likely to have CAs than full term babies (Table 4). Similar results have been reported regarding sex, birth weight for live births and gestational age in Saudi Arabia [*15*]. Additionally, our results are in accordance with the international recommendation to study the complex etiology of low birth weight, which is an important risk factor for CA [*21*].

Conclusions and recommendations

Our observed incidence rate of births with CAs was comparable to or less than the global figure. This is due to under-ascertainment of malformed cases resulting from incomplete cooperation between the National Congenital Anomalies Register and some emirates and lack of post mortem examination of stillbirths and neonatal deaths. Ascertainment could be increased using multiple-source case finding e.g. birth certificates, hospital activity analysis register, perinatal death certificates, and records from a national neonatal screening programme, genetic clinics, and cytogenetic and biochemical genetic diagnostic laboratories.

In addition, for completeness and accuracy, we must compare the number of birth defects recorded in the birth register or hospital register with the number collected through the National Congenital Anomalies Register.

Further efforts are recommended to enhance awareness in health professionals, especially in medical districts where under-reporting is an obvious concern.

Primary preventive programmes should be initiated to reduce congenital malformations, particularly those related to low birth weight and prematurity.

Primary preventive measures for birth defects have to be taken into consideration first because they because they are cheaper and more effective than treatment. They act on risk factors, which are numerous and complex. Preventive measures include treatment of maternal illness e.g. diabetes; rubella vaccination before pregnancy; avoiding medication except when absolute-

ly necessary (under medical supervision); avoiding smoking, both active and passive; good nutrition of pregnant women; periconceptional folic acid supplementation (0.4 mg/day for the whole population, 4.0 mg/day for high risk families); and also extreme maternal age to be avoided through family planning.

There is a necessity to develop health education strategies e.g. premarital counselling, and to enhance screening and diagnostic procedures during the perinatal period.

It is important to evaluate the National Congenital Anomalies Register and to link to international CA register programmes like the International Clearinghouse for Birth Defects Monitoring System (ICBDMS) and the European Registration of Congenital Anomalies and Twins (EUROCAT).

Finally we wish to say that the performance of the National Congenital Anomalies Register in the United Arab Emirates is steadily improving, and we hope to reach international standard in the near future

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