Prevalence of isolated minor congenital anomalies in a regional hospital in Oman

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ABSTRACT

Objective: To study the prevalence of isolated minor congenital anomalies in a regional hospital in Oman.

Methods: We determined the prevalence of isolated minor congenital anomalies in 21988 births during a 10-year period from January 1993 through December 2002 by using data from the hospital-based congenital anomaly register at Nizwa Hospital, Oman.

Results: The total prevalence of congenital anomalies was 37 per 1000 births and that of minor anomalies 12.4 per 1000 births. Hypospadias, talipes deformity and polydactyly were the most common minor anomalies. A nurse or the house resident detected most of the minor anomalies soon after birth during routine neonatal examination.

Conclusion: The study demonstrates the important contribution of minor congenital anomalies to the total prevalence of congenital anomalies. An accurate estimation of the prevalence rates of isolated minor anomalies should be possible as they are easily identifiable with minimal expertise. Case classification of congenital anomalies is important so that case groups are homogeneous and more comparable.

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Congenital anomalies, whether isolated or as components of syndromes, are a common cause of medical intervention, long term disability and death, and pose a significant public health problem.[1] We can divide these into 3 groups: lethal, if the anomaly causes stillbirth or infant death in more than 50% of cases; severe, if we need medical intervention to avert handicap or death; mild (minor) if the anomaly requires medical intervention but outcome and life expectancy are good.[2] Lethal and severe defects together constitute major congenital anomalies. Many newborn infants may have one or more minor anomalies without an associated major malformation.[3] Most epidemiological studies of congenital anomalies including those in the Eastern Mediterranean Region (EMR) have focussed on major defects,[4-6] and epidemiological data on minor anomalies are limited[7,8] because birth defects monitoring systems seldom include these unless major defects accompany them. One reason for this could be that there is no standard classification for the ascertainment of minor anomalies.[9] Several studies have used somewhat arbitrary lists of minor anomalies for exclusion necessitating clinical judgment in their application.[10] Also, the health implications and the morbidity profile of infants with minor anomalies may be underestimated. Approximately 20% of infants with 3 or more anomalies have an associated major malformation.[3] The purpose of the present study is to estimate the prevalence of isolated minor congenital anomalies at Nizwa Hospital, Oman.

Methods. Nizwa Hospital is a major hospital in the Al-Dakhliya region (population 265,000) of Oman. Most of the population in this region comprises Arab Muslims with a high consanguinity

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rate.[11,12] Recording of congenital anomalies in a congenital anomaly register was initiated at the hospital in 1993. This hospital-based register covers all births (live births + still births) at Nizwa Hospital and records all anomalies detected within one week of birth. The diagnosis of a congenital anomaly is based on clinical assessment by a neonatal house resident and validated by a senior pediatrician. Relevant investigations and a review of standard dysmorphology texts[13,14] and the London Dysmorphology Database (LDDB)[15] aid in arriving at an accurate diagnosis. Categorization of anomalies into lethal, severe and minor forms is based on the classification proposed by Czeizel et al.[2] A short report on the contribution of lethal malformations to perinatal mortality[16] has been previously published. Also, a report on major congenital malformations[17] from January 1993 through December 2002 was published. This report includes only minor congenital anomalies and their prevalence and health consequences. The total prevalence of minor congenital anomalies was calculated by dividing the numerator (registered cases of minor anomalies) by the relevant denominator (total live and still births). Only isolated minor anomalies were listed; if they formed part of a multiple malformation syndrome, they were excluded. Care was taken to exclude developmental variants being categorized as minor anomalies. Metabolic disorders presenting in the neonatal period and congenital dislocation of hip were not included in this study.

Results. Eight hundred and fourteen congenital anomalies were detected among 21988 births during the 10-year period from January 1993 through December 2002 giving a prevalence rate of 37 per 1000 births. Of the 814 anomalies, 185 were lethal,[16,17] 356 severe[17] and 273 were minor (mild) abnormalities. The demographic profile of the study cohort is shown in **Table 1**. Of the 273 minor anomalies in the study, 185 occurred in males and 88 in females. However, if 98 urogenital anomalies that occurred exclusively in males are excluded, gender prevalence is almost similar. Minor anomalies were almost equally distributed in relation to maternal age and parity except that talipes was more common in babies of mothers with low parity. Of the neonates with minor anomalies, 88% were term babies and only 14% had low birthweight, which was similar to the hospital low birthweight incidence during the study period. The distribution and frequency of minor anomalies are shown in **Table 2**. The minor anomalies were broadly categorized in 4 groups: urogenital anomalies, musculoskeletal anomalies, cutaneous anomalies and minor anomalies involving head and face. Hypospadias was the most common minor anomaly noted in the study. Of the 73 cases of hypospadias, 70 were glandular or coronal, 3 penile, and none were perineal or penoscrotal. The prevalence of hypospadias was 3.31 per 1000 births and an increased prevalence was noted during the 5-year period between 1998 and 2002 as compared to that between 1993 and 1997. Undescended testis was listed as a minor anomaly only in term infants if testicular descent did not occur by 3 months of age. Of the 25 cases of undescended testis, 9 were bilateral and 16 were unilateral. Of the 52 cases of talipes deformity, 15 were severe and required orthopedic operative intervention and hence were categorized under major malformations. Of the remaining 37 cases, 23 were unilateral. Varus deformity was seen in 27 cases and valgus deformity in 10. Of the 53 cases of polydactyly, all but 3 were cases of postaxial polydactyly, of which 20 were of type A and 30 of type B.[18] The distribution of polydactyly was as follows: 23 cases involved both hands, 16 cases only one hand, 4 cases both hands and feet, 8 cases both feet and 2 cases only one foot. Reliable information on family history was available in 47 cases of polydactyly. Of these, 13 (28%) cases had a family history of polydactyly. Three cases of polydactyly type A were later found to be associated with Laurence Moon Biedl syndrome. Hemangioma was the most common cutaneous anomaly. Five cases had cavernous type of hemangioma, 2 of whom later developed Kasabach-Merritt Phenomenon. Micrognathia without Pierre Robin sequence was noted in 8 cases and was the most common head and neck anomaly, which however showed improvement in early infancy. Six cases of congenital teeth that caused problems in breast-feeding required extraction, after which the natal teeth ceased to be a problem. Since follow-up data was not available in many cases, the exact outcome and the number of cases leading to significant morbidity are not known. **Table 3** provides a comparison of prevalence rates of minor anomalies among studies in the EMR.

Discussion. External minor anomalies usually occur in body parts with complex and variable features such as the face and distal extremities.[13] It is helpful to obtain a family history and to determine whether the anomaly is frequent in the patient's ethnic group before attributing significance to the minor anomaly;[1] however, distinguishing familial features from clinically significant minor anomalies may not always be easy.[19] Prevalence studies of congenital anomalies are useful for establishing baseline rates, for monitoring prevalence trends over time, for identifying clues to etiology and for planning and evaluating health-care services.[9] Reported prevalence rates of minor anomalies vary depending on the definition and diagnostic criteria (inclusion and exclusion criteria) used, the end point of the detection period, the skills and training of the

Table 1 - Demographic profile of neonates with minor congenital anomalies.

Demographic profile	n	(%)
Total number of births (N=21988)		
Omani births	20828	(94.7)
Non Omani births	1160	(5.3)
Total number of minor anomalies(N=273)		
Omani	262	(95.6)
Non Omani	11	(4.4)
Gender		
Males	185	(67.8)
Females	88	(32.2)
Gestational age		
Term	240	(88)
Preterm	33	(12)
Birth weight		
≥2500 g	237	(86)
<2500 g	38	(14)

Table 3 - Comparison of prevalence of minor anomalies among studies in the Eastern Mediterranean Region.

Anomalies	Present study	Mir et al[7]	Refat et al[6]
Study period	1993-2002	1982-1984	1411H-1413H
Total births	21988*	32332†	30159†
Prevalence of congenital anomalies/1000 births	37	23.8	22.7
Hypospadias	3.31	1.54	0.7
Talipes	2.36	3.83	1.0
Polydactyly	2.41	1.54	1.1
Syndactyly	0.36	0.12	0.5
Cryptorchidism	1.14	-	-
Cutaneous abnormalities	2.27	2.59	0.5
Hemangiomas	0.73	0.99	

*Total live + still births;
†Total live births

Table 2 - Prevalence and gender distribution of minor congenital anomalies.

Type of anomaly	n	Prevalence 1000 births	Gender distribution	
			Male	Female
Urogenital				
Undescended testis	25	1.14	25	-
Hypospadias	73	3.31	73	-
Subtotal	98		98	-
Musculoskeletal				
Talipes deformity	37 *	2.36†	17	20
Polydactyly	53	2.41	24	29
Syndactyly	07	0.36	5	02
Deformities of head, face and chest	09	0.41	4	05
Subtotal	106		50	56
Cutaneous				
Hemangiomas	16	0.73	12	04
Melanocytic naevi	05	0.22	3	02
Ichthyosis	09	0.40	5	04
Preauricular tags	13	0.59	6	07
Skin tags	07	0.12	4	03
Subtotal	50	2.27	30	20
Head and face				
Microglossia	01	0.05	-	01
Macroglossia	03	0.14	1	02
Micrognathia	08	0.36	4	04
Congenital tooth/teeth	06	0.27	2	04
Miscellaneous	01	0.05	-	01
Subtotal	19		7	12
Total	**273**	**12.41**	**185**	**88**

*15 cases of severe talipes excluded and only 37 listed as minor anomaly
†prevalence of talipes includes all 52 cases (37 minor + 15 major) for comparison

examiner and the methods of case ascertainment including data collection, sources of notification and type of surveillance system.[17,20] These may prevent meaningful comparison among studies. The total prevalence of congenital anomalies in the present study is similar to the Scottish prevalence of congenital anomalies (324 per 10,000 births),[21] but higher than 238 per 10,000 live births and 216.8 per 10,000 births reported by Mir et al[7] in Benghazi, Libya and the EUROCAT registries (1993-2002)[22] respectively. Some degree of selection bias was probably in effect because we conducted our study in a major hospital in a region of Oman which also receives referrals of high-risk pregnancies and this may partially explain the higher prevalence in comparison with other studies. Also, EUROCAT has a list of anomalies which are not to be transmitted to the Central Registry and are excluded from registration.[23]

Hypospadias was the most common minor anomaly noted in the present study. Distal hypospadias accounts for more than three-quarters of the cases of hypospadias, as also observed in this study and is thus, more common than the severe forms.[1] The prevalence rate in our study is higher than that reported from Libya by Mir et al[7] (1.54 per 1000) during a study period from July 1982 to June 1984, but lower than that reported by Ahmed et al[24] (4.1 per 1000) from Glasgow, Scotland. In the 1970s and 1980s, some European registries reported an upward trend in the birth prevalence of hypospadias.[25] Paulozzi et al[26] also observed an approximate doubling of hypospadias rates in the United States during the same period; also, the rate of severe cases increased while the ratio of mild to severe cases decreased. However, a study which examined more recent data from countries participating in the International Clearinghouse for Birth Defects Monitoring Systems (ICBDMS) to address the questions of whether such increases are worldwide and continuing found that the increases leveled off in many systems after 1985, and increases were not seen in less affluent nations.[27] Epidemiological data on hypospadias is very limited from countries of the EMR and we require more data to study trends in the rates of hypospadias in this region. A recent prospective study in a university hospital in Finland[28] reported only one case of scrotal hypospadias and all other cases were glanular or coronal. The EUROCAT guidelines exclude registration of first-degree hypospadias unless associated with other specified major anomalies.[23] A recent report showed that registers covering a large number of hospitals and clinicians had difficulty accurately excluding glanular cases due to different interpretations of the exclusion guideline thereby producing inconsistent, inaccurate or incomplete reporting of hypospadias.[29] It suggested an interrelation between the prevalence of hypospadias, proportion of glanular or coronal cases, and proportion of cases undergoing surgery, which must be interpreted together, and that all cases of hypospadias need to be reported irrespective of location.[29] Frydman et al[30] reported uncomplicated hypospadias in several members of a large consanguineous family, and we need further studies in populations of this region with a high rate of consanguinity. Complete prepuce may cover a hypospadias on the glans,[31] and awareness of the anomaly is necessary for early detection and surgical treatment, especially in societies which have ritual circumcision in the neonatal period, because the foreskin is required for hypospadias repair. Cryptorchidism is the most frequent developmental abnormality in boys, present in >1% of infants above 3 months of age and in 0.8% at one year of age.[32] A search for the cause is often disappointing except in bilateral cases or associated malformations.[32] Brucker-Davis et al[32] suggested a possible in utero impact of hormonally active environmental factors such as pesticides with estrogenic and antiandrogenic effect. Studies on subsegments of the population with maximal exposure potential need to be conducted.[33,34] Musculoskeletal system anomalies accounted for more than one third of the minor anomalies; talipes deformity and polydactyly were the most common anomalies within the group. The birth prevalence of clubfoot (equinovarus deformity) varies among different ethnic groups. The recurrence risk for subsequent offspring of normal parents with an affected child is approximately 2-3%, but 20-30% for offspring of involved parents.[1] Though polydactyly is one of the most common congenital anomalies, we do not know its true prevalence due to the paucity of epidemiological data. It may occur in isolation or in association with other congenital anomalies.[35] Postaxial polydactyly of the hand is the most frequent type. Reported risk factors are African black ethnicity, male gender, twinning, low maternal education, parental consanguinity and a positive family history.[36,37] The recorded prevalence of hemangiomas in this series may be an underestimate as approximately 55% are present at birth and the remainder develop in the first weeks of life.[1]

Epidemiological study of congenital anomalies is difficult as individual defects are rare, they are etiologically heterogeneous and the causes of most remain unknown and the defects identified at birth represent only the birth prevalence and not the true incidence of the condition.[38] Isolated birth defects may have different epidemiological characteristics and risk factors and thus different implications for prognosis and recurrence risks from those that occur in combination with other defects.[1,39,40] Hence, standardization in definition and classification of birth defects is important for comparison among

studies and identification of risk factors. The magnitude of disease risk and thereby prevalence of anomalies varies in different populations reflecting a complex interaction of genetic variation and environmental factors. This study provides the prevalence of isolated minor anomalies in a region of Oman. However, there is a need to conduct further studies of these anomalies in other regions of Oman to explore differences in prevalence and identify risk factors including exposure to a teratogen. We need to develop a specific protocol of minor anomalies, as this may serve as a reminder and result in greater awareness and early identification of minor anomalies by the physician.[3] Marden et al[41] reported that 14% of newborn babies have a single minor anomaly detectable by surface examination (except for dermatoglyphics), 0.8% have 2 minor anomalies and 0.5% have ≥3 anomalies.[13] The frequency of major malformations was not increased in the first subgroup. However, in the second subgroup, the frequency of a major defect was 5 times that of the general group, and 90% of babies in the third subgroup had one or more major defects. Further studies by Mehes et al[42] and Leppig et al[3] demonstrated that 26% and 19.6% of newborn babies with ≥3 anomalies respectively have a major defect, a much lower incidence than that documented by Marden et al.[41]

A physician examining newborn infants should be familiar with minor anomalies because they are common and serve as valuable clues in the detection of congenital disorders.[3,43] Also, because of psychological implications,[44] screening all neonates for minor anomalies is important for their early detection and intervention.

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