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Pattern of Major Congenital Anomalies in Southwestern Saudi Arabia

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Background: Currently, in the Arabian Peninsula, genetically determined disorders account for an increasing proportion of death, morbidity, chronic handicap, and disability

Aim: To study the pattern and classification of MCAs in Asir region, during six-year period, in order to allow proper genetic counseling, early management and rehabilitation.

Method: The study included all neonates with congenital anomalies referred to Asir Central Hospital from 1997 to 2002. Cases with genetic syndromes were diagnosed by review of Mandelian inheritance in man and the London dysmorphology database. The major congenital anomalies were classified according to the ICD-10 system, and multiple MCAs were counted only once by the system of the most major anomaly

Results: Of 1171 newborns admitted to neonatal intensive care unit (NICU) at Asir Central Hospital, 691 newborns were proved to have congenital anomalies, constituting 59.1% of all admissions. According to ICD-10 classification of congenital anomalies, the systems involved in the MCAs investigated were (in descending order of frequency) as follows: digestive 28.6%, central nervous 26.1%, circulatory 16.5%, urogenital 7.1%, face and neck 4.1%, respiratory 6.2%, musculoskeletal 3.6%, chromosomal 3.3%, and other anomalies 4.5%.

Conclusion: Congenital anomalies represent the main reason for referral to NICU in Asir region, and this implies that congenital malformations constitute significantly to perinatal and infant morbidity in the region. Premarital counseling should be advised, especially in the presence of parental consanguinity and family history of a congenitally malformed child.

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Congenital disorders present at birth in 2 to 4% of all neonates[1]. These abnormalities may be caused by environmental factors that affect the mother, or are inherited via abnormal genes from the carrier or affected parent. Such disorders account for about 20% of deaths during the neonatal period and a higher percentage of morbidity in infancy and childhood[2]. With the dramatic decreases in infant mortality due to improvement in the control of infections and malnutrition-related disorders, chronic disabling conditions are an emerging challenge facing developing and industrialized nations. In Saudi Arabia, the incidence of major congenital anomalies accounted for 22.7 per 1000 live births[3].

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The Asir region is an area of about 80,000 Km^2, with a population of about 1.2 million people. Asir Central Hospital is the main referral hospital, and serves as the teaching hospital of the medical school in the region. All complicated and terminal cases from the 17 hospital in the region are routinely referred to this hospital. As such, neonates with major congenital anomalies (MCAs) referred to the hospital provide a fair reflection of such conditions in this area.

The aim is to study the pattern and classification of MCAs in Asir region, in a six-year period, in order to allow proper genetic counseling, early management and rehabilitation.

METHODS

The study included all neonates with congenital anomalies referred to Asir Central Hospital from 1997 to 2002. All neonates identified with congenital anomalies were admitted to NICU for observation, investigation, evaluation and management. They were examined to identify congenital defects. Photographs, radiographs, necropsy reports and chromosomal studies were included when recommended.

Cases with genetic syndromes were diagnosed by review of *Mandelian inheritance in man* and the *London dysmorphology database*[4,5]. The major congenital anomalies were classified according to the ICD-10 system, and multiple MCAs were counted only once by the system of the most major anomaly[6].

RESULTS

Table 1 shows that out of 1171 newborns admitted to neonatal intensive care unit (NICU), 691 newborns were proved to have congenital anomalies, constituting 59.1% of all admissions. These congenital anomaly cases constituted significantly higher proportion of male admissions than its counterpart of female admissions (X^2= 18.34, $P<0.001$).

Table 1. Number (%) of admissions in the neonatal unit at Asir Central Hospital within six-year period (1997-2002) and the corresponding number of congenital anomaly cases.

Sex	Total no. admissions	Newborns with anomalies	
		No.	%
Male	581	384	66.1
Female	589	317	53.8
Total	1170	691	59.1

According to ICD-10 classification of congenital anomalies (Table 2), the systems involved in the MCAs investigated were (in descending order of frequency) as follows: digestive (28.6%), central nervous (26.1%), circulatory (16.5%), urogenital (7.1%), face and neck (4.1%), respiratory (6.2%), musculoskeletal (3.6%), chromosomal (3.3%), and other anomalies (4.5%).

Table 3 shows the rank order of different MCAs in the present study in comparison with other studies. Digestive system anomaly ranked first in the present study, while it was forth in both Saudi Arabian and Libyan studies. On the other hand, while circulatory system anomaly ranked first in both Saudi Arabia and UAE, it ranked 3rd in the present study.

Table 2. Frequency of major congenital anomalies (MCAs) in Asir region by system according to ICD-10 classification

MCAs by system (ICD-10)	No.	%	% of total
Q00-Q07 Central nervous system			
spina bifida and encephaloceles	81	45.0	
hydrocephalus without spinabifida	55	30.6	
microcephaly	30	16.7	
crynosyenstosis	6	3.3	
porencephaly	5	2.8	
anencephaly	3	1.6	
Subtotal	180	100	26.1
Q10-Q18 Eye,Ear,face and neck	28	100	4.1
Q20-Q28 Circulatory system			
ventricular septal defect	60	52.6	
hypoplastic single ventricle	14	12.3	
tetralogy of fallot	13	11.4	
endocardial cushion defects	12	10.5	
transposition of great vessels	8	7.0	
complex cardiac anomalies	7	6.2	
Subtotal	114	100	16.5
Q30-Q34 Respiratory system			
- tracheo-oesophageal fistula	43	100	6.2
Q35-Q37 Cleft lip and Cleft palate	25	100	3.6
Q38-Q45 Other digestive system			
intestinal obstruction	56	32.4	
imperforated anus	40	23.1	
Hisrshsprung disease	39	22.5	
Diaphragmatic hernia	38	22.0	
Subtotal	173	100	25.0
Q50-Q64 Urogenital system			
posterior urethral valve	10	20.4	
undescended testes	10	20.4	
hypospadius	9	18.4	
ambigeous genitalia	9	18.4	
polycystic kidney	8	16.3	
Poter syndrome	3	6.1	
Subtotal	49	100	7.1
Q65-Q79 Musculoskeletal system	25	100	3.6
Q80-Q89 Other anomalies			
Cutaneous	15	48.4	
hydrocele	11	35.5	
tumor cyst	5	16.1	
Subtotal	31	100	4.5
Q90-Q99 Chromosomal (not classified elsewhere)			
trisomy 21	15	62.2	
trisomy 18	5	21.7	
trisomy 13	3	13.1	
Subtotal	23	100	3.3
TOTAL	691		100

Table 3. Rank order of MCAs in the present study in comparison with other studies in different countries[7]

System involved	Saudi Arabia	Libya	UAE	Nijeria	current study
Digestive	4	4	-	-	1
Central nervous	3	-	-	2	2
Circulatory	1	2	1	-	3
Musculoskeletal	2	1	2	1	5
Chromosomal	4	3	-	-	6

NB. table represent the data available.

DISCUSSION

The social and economic conditions in the Arabian Peninsula have improved enormously in the last 3 decades. This led to the sharp decline in the incidence of infectious diseases and diseases related to malnutrition. Currently genetically determined disorders account for an increasing proportion of death, morbidity, chronic handicap and disability[8]. During 1985-1989, 19% of pediatric inpatients - in King Khalid University Hospital in Riyad city of Saudi Arabia – had congenital or genetically-determined disorders[9]. However, in the present study, neonates with MCAs constituted 59.1% of all neonates referred to NICU of Asir Central Hospital during six years.

This high frequency of MCAs might have resulted from common consanguineous marriages, which led to the preservation of rare mutations kept in a genetically homogenous population. Several publications indicate that consanguineous marriages in Saudi Arabia are high (60%) and this has provided a background in which these genetic diseases abound[10,11]. This implies that congenital malformations constitute significantly 59.1% to perinatal and infant morbidity in Asir region.

More male admissions than females with MCAs were noted in the present study. Male preponderance concurs with the findings of other studies[12-14]. It may be speculated that either the females were afflicted with more lethal congenital malformations and could not survive to be referred or that their malformations were too mild to be referred.

Congenital anomalies in the present study were classified according to the ICD-10 classification and multiple anomalies were counted only once based on the system involved in the most major malformation. It is possible that some cases of congenital anomalies could have been undetected, particularly in asymptomatic neonates, in cases of internal anomalies and perinatal deaths. It is unlikely, however, that any major, external or obvious malformations were missed. Digestive system anomalies ranked first in the present study, although in other studies it showed lower rates[7]. These low rates were attributed to the routine ultrasonography in the neonatal intensive care unit. However, in this study, ultrasonography is routinely done to all referred cases with suspected congenital anomaly. The most frequent lesions of this system were intestinal obstruction, tracheoesophageal fistula, imperforate anus and diaphragmatic hernia. This finding concurs with the results of Al-Qassim study in the central region of Saudi Arabia[15].

Central nervous system anomalies ranked second in the present study. However, spina bifida and encephalocele together ranked first among all reported congenital malformations. In the

developed countries, following mass promotion and mandatory prescription of folic acid for pregnant women, the incidence of neural tube defect has markedly declined[14,16,17].

Throughout the world, chromosomal aberrations are among the least defined causes of congenital anomalies[18]. This was in concordance with the results of the present study, where these anomalies ranked six in frequency. However, trisomy 21 constituted two-thirds of all these chromosomal anomalies. This syndrome is a major cause of mental retardation, congenital heart problems and gastrointestinal malformations[19].

CONCLUSION

Congenital anomalies represent the main reason for referral to NICU in Asir region, and this implies that congenital malformations play a major role in perinatal and infant morbidity in the region. Our results probably underestimate the number of congenital anomalies in Asir region as a result of possible non-referral of minor anomalies, in addition to non-inclusion of stillbirths and neonatal deaths. However, the results of the present study aim at addressing the categories of MCAs that might bring the burden of the future handicapping conditions with their social and economical consequences.

Accurate and early diagnosis of congenital malformations is the key to proper management of cases. Premarital counseling is advised, especially in the presence of parental consanguinity and family history of a congenitally malformed child. Because of the high frequency rate of neural tube defects, proper prenatal diagnosis is recommended. However, cultural and religious factors are to be considered. If prevention is not possible, it becomes even more crucial to anticipate and plan accordingly for the management.

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