Pattern of congenital anomalies among newborns, infants and children in Aden city

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Abstract

The field of dysmorphology has expanded dramatically as the number of recognizable patterns of malformation has more than tripled during the last 30 years. Major congenital anomalies are currently the leading cause of perinatal, neonatal and infant mortality worldwide, including Yemen. A basic method to investigate congenital anomalies is through medical review records at hospitals and child maternity services.

A retrospective analytic study through review of medical records was conducted includeding 1920 patients in all major congenital anomalies with a male to female ratio of 2:1 within age range from 1 day to15 years at Al-Sadaqa and Al-Gamhouria Teaching Hospitals, Aden city, Yemen, during January 2000 to December 2007. Digestive system (DTS) 649 (33.8%) formed the commonest major congenital anomalies, followed by circulatory system (CVS) 416 (21.7%), central nervous system (CNS) 273 (14.2%), urogenital system (UGS) 202 (10.5%) and musculoskeletal system (MSK) 137 (7.1%).

Generation of the available information will form the basis to reflect the magnitude of these birth defects, their pattern and any associated risk factors. These results will have important implications in planning appropriate preventive, therapeutic and rehabilitative programs. Future plan would include the implementation of innovative health education strategies and standard screening with sophisticated diagnostic procedures.

Keywords: Congenital anomalies, digestive system, circulatory system, central nervous system, urogenital system and musculoskeletal system.

Introduction:

Congenital anomaly has been defined according to the World Health Organization as any morphological, functional, biochemical or molecular defects that may develop in the embryo and fetus at the time of conception until birth, and maybe detected at birth or later [26]. It is estimated that 3 million fetuses and infants are born each year with major malformations [10]. Global estimates suggest that congenital anomalies affect 2–3% of births [12]. Approximately, around 303 000 newborns worldwide die within 4 weeks of birth annually due to congenital anomalies which contribute to long-term disability having important impacts on patients, families, health-care systems, and societies [29].

These birth defects have tripled during the last 30 years. Major congenital anomalies have a higher incidence in Arabs of Gulf region with multi-regional variations in rate and pattern with multifactorial etiologies. Although congenital anomalies may be the result of one or more genetic, infectious, nutritional or environmental factors, it is often difficult to identify the exact causes [29, 27]. It has been reported that more than 40% are idiopathic and have a high prevalence rate in consanguineous marriages [17, 3].

Major congenital anomalies are the leading causes of perinatal, infant morbidity and mortality in industrialized, developing and underdeveloped countries, including Yemen [28]. Congenital anomalies constitute the fifth largest cause, being responsible for an estimated 9% of neonatal deaths [16]. The most common and severe congenital anomalies are heart defects, neural tube

defects and Down syndrome. It is apparent that birth defects are a major public health problem because of their significant contribution to mortality and morbidity with future handicaps [13].

The etiology of congenital anomalies is categorized with 25% of them being caused by a chromosomal anomaly; 20% caused by a single gene disorder; 5% related to an environmental factor and approximately 50% are caused by multiple factors [9]

The role of pediatricians, dealing with birth defects, is to give a diagnostic opinion, to help understand the etiology and to discuss genetic aspects of condition, to advise investigation pertinent to diagnosis and to advise on prognosis and therapeutic options, as well as to discuss the risk of recurrence in pregnancy and if prenatal testing is available.

The main objectives of this study was to reflect the magnitude of the birth defects, their pattern, sex distribution in neonate, infants and children and to classify the major congenital anomalies according to the ICD-10 system by sex distribution to major congenital anomalies and to come out with future recommendations.

Patients and methods:

The design of this research was a descriptive analytic study through review of medical records with collection and interpretation of data that included samples of 1920 patients of both sexes within the age ranging from 1 day to15 years, who were enrolled in this study. The setting of the study was at Al-Wahda and Al-Gamhouria Teaching Hospitals, Aden city during the period of January 2000 to December 2007.

The inclusion criteria were all Yemeni patients with congenital anomalies in any system of the body which affected their survival or caused structural, cosmetic or functional handicaps and who required medical or surgical intervention. Major congenital anomalies (MCAs) were counted only once by system of most major anomaly.

The research approval was taken by the ethical committee at the Faculty of Medicine and Health Sciences, University of Aden, Yemen.

Results:

A total of 1920 patients of both sexes were analyzed where males were 1260 (65.7%) predominating females 660 (34.3%) in all major congenital anomalies, with a male to female ratio of 2:1. Digestive system (DTS) 649 (33.8%) formed the commonest major congenital anomalies, followed by the circulatory system (CVS) 416 (21.7%), central nervous system (CNS) 273 (14.2%), urogenital system (UGS) 202 (10.5%) and musculoskeletal system (MSK) 137 (7.1%). Chromosomal aberrations were detected in 87 (4.5%) of the studied patients. (Table 1) (Figure1)

Table.1 International Classification of Statistical Disease Major Congenital Anomalies by Systems (MCAs)

MCAs by System (ICD-10)	**No.**	**%**
Q00-Q07Central nervous system	273	14.2
Q20-Q28 Circulatory	416	21.7
Q30-Q34 Respiratory	28	1.5
Q35-Q37 Cleft lip & Cleft Palate	41	2.2
Q38-Q45 Digestive	649	33.8
Q50-Q64 Urogenital	202	10.5
Q65-Q79 Musculoskeletal	137	7.1
Q90-Q99 Chromosomal aberrations	87	4.5
Q 80-89 Others	87	4.5
Total	1920	100

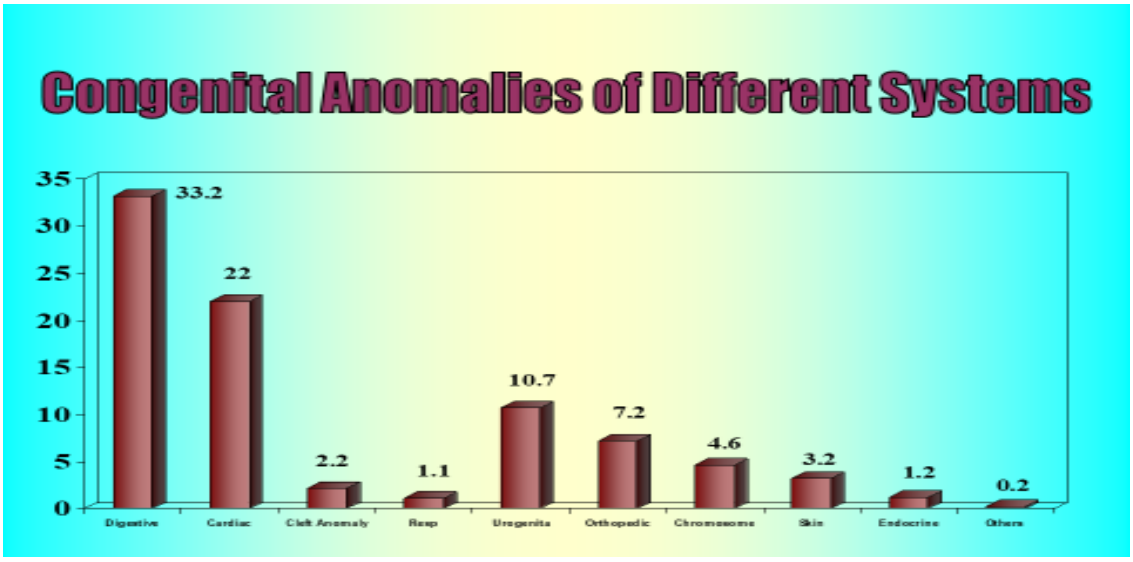

Figure No.1 Congenital anomalies by different systems

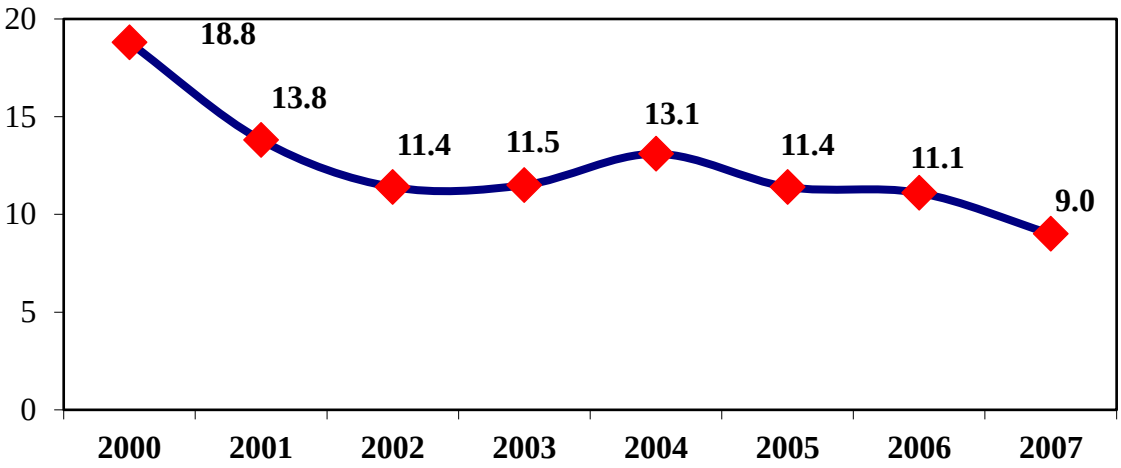

Figure No.2 Congenital anomalies by years

There was a progressive decrease in the number of congenital anomalies throughout the successive years of 2000 to 2003 and, then ,remaining almost constant until 2006 with a further decrease in 2007. (Figure 2)

Table.2 Major Congenital Anomalies of Different Systems By Sex and Defect

Major Congenital Anomalies	Sex		
	Male No (%)	Female No (%)	Total No (%)
Central nervous system	145 (7.5)	128 (6.7)	273 (14.2)
Circulatory	233 (12.1)	183 (9.5)	416 (21.6)
Respiratory	20 (1.1)	8 (0.4)	28 (1.5)
Cleft lip and palate	26 (1.3)	15(0.8)	41 (2.1)
Digestive	472 (24.6)	177 (9.2)	649 (33.8)
Urogenital	193(10)	9 (0.5)	202 (10.5)
Musculoskeletal	83 (4.3)	54 (2.8)	137 (7.1)
Chromosomal	55 (2.9)	32 (1.6)	87 (4.5)
Skin	26 (1.3)	34 (1.8)	60 (3.1)
Endocrine	6 (0.3)	17 (0.9)	23 (1.2)
Others	1 (0.05s)	3 (0.15)	4 (0.2)
Total	1260 (65.6)	660 (34.4)	1920 (100%)

The major congenital anomalies showed a higher male preponderance than females in all the different systems with the exception of the skin and endocrine which showed a higher female predominance. (Table 2)

Table 3.Major Congenital Anomalies of Cardiovascular System (CVS) By Sex and Defect

Cardiac defects	Sex		
	Male No (%)	Female No (%)	Total No (%)
VSD	71 (17.1)	60 (14.4)	131(31.5)
PDA	28 (6.7)	24 (5.8)	52 (12.5)
ASD	23 (5.5)	25 (6.0)	48 (11.5)
ECD	7 (1.7)	5 (1.2)	12 (2.9)
PS	5 (1.2)	4 (1.0)	9 (2.2)
TOF	5 (1.2)	4 (1.0)	9 (2.2)
TGA	19 (4.5)	4 (1.0)	23 (5.5)
Dextrocardia	6 (1.4)	2(0.5)	8 (1.9)
Others	69 (16.6)	55 (13.2)	124 (29.8)
Total	233 (56)	183 (44)	416 (100)

All the cardiac defects were more common in males than females ,except the ASD which showed a female predominance. (Table 3)

Table 4. Major Congenital Anomalies of Central Nervous System (CNS) By Sex and Defect

Central Nervous System	Sex		Total
	Male No. (%)	Female No. (%)	No. (%)
Hydrocpehalus	67 (25.5)	68 (25.8)	135 (51.3)
Meningocele	59 (22.4)	46 (17.5)	105 (39.9)
Anencephaly	4 (1.4)	1 (0.4)	5 (1.8)
Eencephalocele	1 (0.4)	3 (1.2)	4 (1.6)
Others	7 (2.7)	7 (2.7)	14 (5.4)
Total	138 (52.5)	125 (47.5)	263 (100)

Figure 3 CNS anomalies in the newborns

Among CNS anomalies, hydrocephalus was equal in both sexes, males 67 (25.5%) versus female 68 (25.8) and meningocele was higher in males 59 (22.4%) than in females 46 (17.5%). (Table 4) (Figure 3)

Table 5. Major Congenital Anomalies of Digestive System By Sex and Defect			
Digestive System	Sex		Total
	Male No. (%)	Female No. (%)	No. (%)
TEF	19 (2.9)	9 (1.4)	28 (4.3)
Congenital hypertrophic pyloric stenosis	54 (8.3)	11 (1.7)	65 (10)
Diaphragmatic hernia	5 (0.8)	1 (0.1)	6 (0.9)
Rectoanal anomalies	99 (15.3)	70 (10.8)	169 (26.1)
Umbilical hernia	16 (2.5)	16 (2.5)	32 (5.0)
Inguinal hernia	214 (33)	48 (7.4)	262 (40.4)
Megacolon	35 (5.4)	12 (1.8)	47 (7.2)
Duodenal stenosis	12 (1.8)	2 (0.3)	14 (2.1)
Tongue tie	18 (2.8)	7 (1.1)	25 (3.9)
Total	472 (72.8)	176 (27.2)	648 (100)

The most common major congenital anomalies in the digestive system were inguinal hernias 262 (40.4%), rectoanal anomalies 169 ((26.1%) followed by congenital hypertrophic pyloric stenosis 65 (10%) which showed higher percentage in males than females, except for umbilical hernia which was the same for both sexes 16 (2.5%). (Table 5) and (Figure 4)

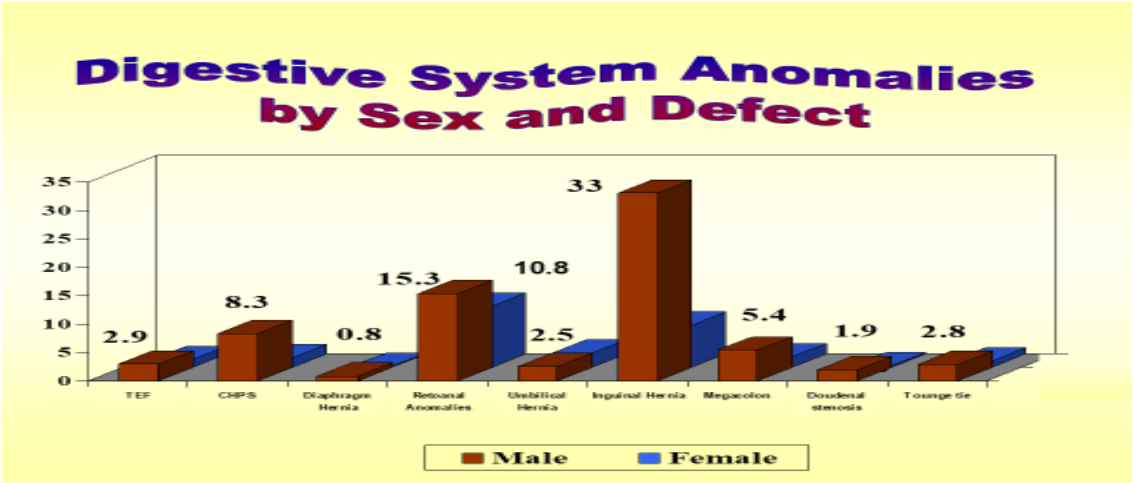

Figure 4 Digestive system anomalies by sex and defect

Undescended testes 64 (33.2%) was the most common congenital anomaly of the urogenital system, followed by hypospadias 56 (29.2%) and hydrocele 42 (21.8%). (Table 7) (Figure 5).

Table 7. Major Congenital Anomalies of Urogenital System By Defect	
Urogenital System	**Total No (%)**
Undescended Testis	64 (33.2)
Hypospadias	56 (29.2)
Hydrocele	42 (21.8)
Bladder Exostrophy	2 (1.5)
Posterior Urethral Valves	2 (1.5)
Epispadias	2 (1.5)
Others	17 (9)
Total	193 (100%)

Figure 5 Urogenital anomalies by sex and age

The musculoskeletal anomalies showed congenital hip dislocation to be the most common anomaly with a higher percentage in males (97.2%), followed by talipes equino varus (TEV) which was more frequent in females (72.8%).

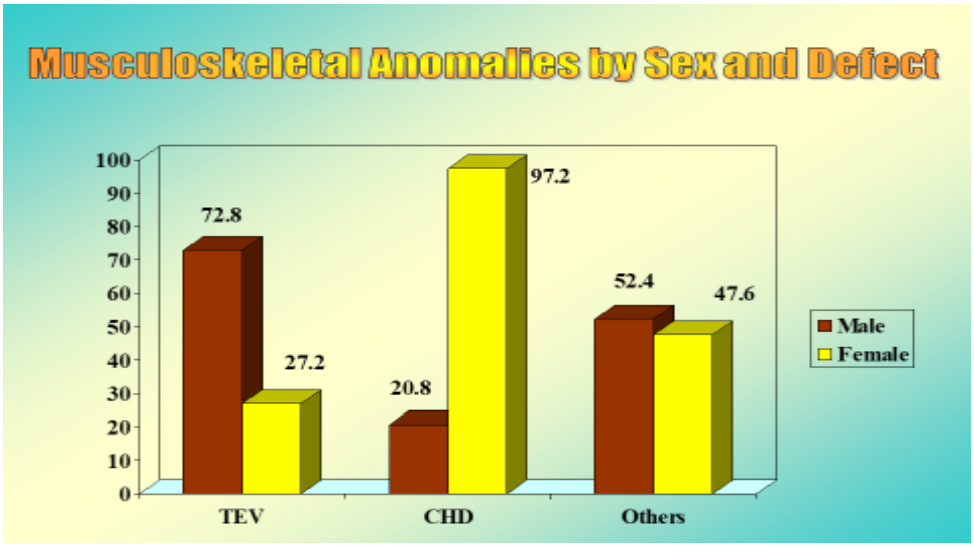

Figure 6 Musculoskeletal anomalies by sex and age

Cleft lip and cleft palate comprised a total of 41 patients with 19 (46.3%) having cleft palate, and 6 (14.6%) cleft lip alone and 16 (39.1%) with both cleft lip and cleft palate.

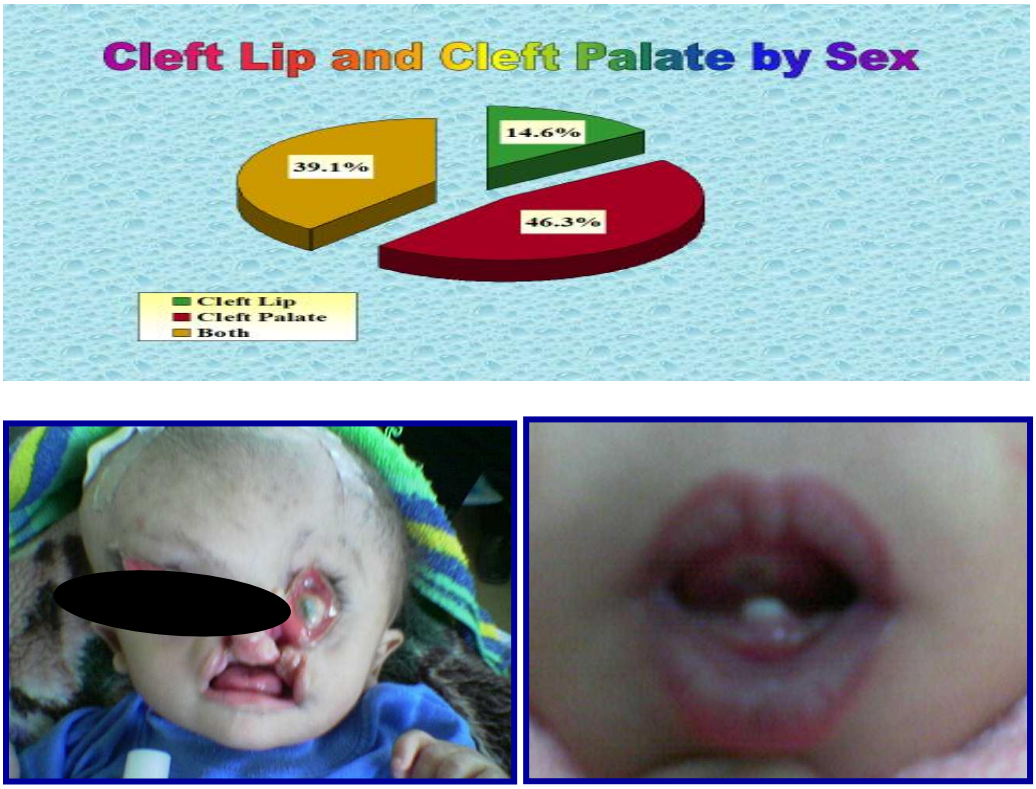

Figure 7 Cleft Lip and Cleft Plate

Chromosomal aberrations formed a total of 87 (4.5%) of birth defects with Down's syndrome comprising 88.5% and Edward's syndrome 4.6% of them.

Figure 8 Chromosomal aberrations by sex and age

Discussion:

In this study, a total of 1920 patients of both sexes had major congenital anomalies with higher male predominance (65.7%) with a male to female ratio of 2:1; in contrast to other reports with higher female frequency [27, 22]. Males outnumbered females in all types of birth defects comparable to many literature reports [11, 7]. The relative sex difference of various malformations might be due to multifactorial factors [11, 22].

A higher significant proportion of major congenital anomalies were detected in DTS (33.8%), followed by CVS (21.7%) in contrast to studies in India and Egypt where MSK and CNS anomalies were the highest; while in Saudi Arabia, the most frequent were UGS anomalies [11, 7,

25, 23]. A systemic review and meta-analysis of congenital anomalies among hospital studies in India showed that CNS anomalies were the most common, followed by the MCKS, while CVS anomalies had the lowest prevalence, similarly reported in Iraq and Egypt [24, 1, 30, 21, 19]. Another report showed that the most common anomalies were detected in the CNS and UGS ,while in a different study it was DTS followed by UGS, MSKS and CVS [14, 5, 8]. It is rather difficult to predict that CNS, anomalies are not common in Yemen, but it may be explained by the fact that they may be missed or underdiagnosed, or patients die before being diagnosed.

Other studies showed that the predominant major congenital anomalies were the MSK (33.2%), followed by DTS (15%). In this study, DTS anomalies were more common than MSK anomalies; with congenital hip dislocation being the most common one in MSK group and cleft lip and cleft palate similar to a study reported in India [24]. The relative difference in the occurrence of various malformations in different reports might be due to geographic, genetic, environmental and racial differences [22, 7, 1, 2].

Finally, in this study, a higher significant proportion of major congenital anomalies were detected in DTS, followed by CVS. Males almost predominated females in all birth defects. VSD was the commonest acyanotic heart defect and TGA the frequent cyanotic one. UGS anomalies ranked the third frequent defect with undescended testes and hypospadias.

Hydrocephaly and menigocele were common CNS defects with no sex difference. Talipes equino varus and congenital hip dislocation were the predominant bone defects with sex predilection, as similarly reported in Iran [2]. Down syndrome comprised the highest percentage of chromosomal aberrations as known globally.

In the literature, it is documented that a link exists between some birth defects and specific environmental risk factors. Currently, many epidemiological and experimental studies are conducted to bring data on the most probable risk factors for single or groups of congenital anomalies. Hence, it is worth mentioning that until this current time, there is no unified monitoring system for birth defects worldwide [6].

Furthermore, the role of team of paediatricians for patients with congenital anomalies is to rapidly recognize various types of congenital anomalies, and to provide basic care, treatment and counselling, followed by referral of the patients to a specialist when necessary [24, 1, 20, 18]. It is extremely important to have a general detailed data on the magnitude of congenital anomalies in the country since some of these conditions can be prevented through the role of primary care interventions in the maternity which should be mainly targeted towards women in the preconception, intra-conception and antenatal periods.

Conclusions:

Congenital anomalies carry a high burden to affected individuals and families in quality of life, community participation and services needed. It is of paramount importance for early diagnosis and appropriate management to help plan future care,to initiate primary preventive and standard screening program, and diagnostic procedures during perinatal period that will possibly reduce morbidity and mortality. This should be followed by the creation of an effective support service of physical, educational, vocational and social rehabilitation for babies surviving with disabilities and handicaps. Future cross-sectional epidemiological studies are urgently required with the establishment of a National Registration Scheme for genetic disorders and congenital anomalies in Aden, Yemen.

Limitations of the study:

This study is a retrospective one which has some limitations. It is difficult to estimate the actual prevalence of birth defects since the diagnosis does not include stillbirth and neonatal, infant and child deaths.

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نمط العيوب الخلقية بين المواليد والرضع والأطفال في مدينة عدن

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الملخص

العيوب الخلقية تزداد انتشاراً بشكل مطرد حبت يفوق ظهور الأنماط المعروفة من هذه التشوهات الثلاثة أضعاف عما كانت عليه خلال السنوات الثلاثين الماضية. تعد العيوب الخلقية الشائعة سببا مًهماً في زيادة الوفيات والإعاقات الدهنية والجسدية خاصة عند حديثي الولادة والرضع في جميع أنحاء العالم عامة وفي اليمن خاصة. ويُتعرف على العيوب الخلقية والزيادة في حدوثها من خلال سجلات المراجعة الطبية في المستشفيات ومراكز رعاية الأمومة والطفولة.

أجريت هذه الدراسة التحليلية بأثر رجعي من خلال مراجعة السجلات الطبية التي شملت 1920 مصابا بمختلف العيوب الخلقية الرئيسية وبنسبة 1:2 بين الذكور إلى الإناث، ضمن الفئة العمرية من 1 إلى 15 سنة في مستشفى الصداقة ومستشفى الجمهورية التعليميين، بمدينة عدن – اليمن، خلال المدة من يناير 2000 إلى ديسمبر 2007. أظهرت النتائج أعلى نسبة من اهم العيوب الخلقية في الجهاز الهضمي 649 (33.8 ٪)، تليها الدورة الدموية 416 (21.7 ٪)، الجهاز العصبي المركزي 273 (14.2 ٪)، الجهاز البولي التناسلي 202 (10.5٪) والجهاز الحركي 137 (7.1٪).

ستكون المعلومات المتاحة أساساً لإظهار حجم هذه العيوب الخلقية ونمطها وأي عوامل خطر مرتبطة بها. ستكون لهذه النتائج آثار مهمة في تخطيط البرامج الوقائية والعلاجية والتأهيلية المناسبة، ستشمل الخطة المستقبلية تنفيذ استراتيجيات مبتكرة للتثقيف الصحي وفحص قياسي مع إجراءات تشخيصية معقدة.

الكلمات المفتاحية: التشوهات الخلقية، الجهاز الهضمي، الدورة الدموية، الجهاز العصبي المركزي، الجهاز البولي التناسلي والجهاز الحركي.