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Congenital Anomalies in Yemeni Newborns: A Retrospective Study at a Tertiary Care Hospital in Sana'a City

Sahar A. Thabet[1], Ahmed S. Hudna[2*], Abdulrahman M. Alhadi[3], Hala Al-Kherbash[3]

[1] *Pediatrics Department, Modern European Hospital, Sana'a, Yemen*

[2] *Pediatrics Department, Faculty of Medicine and Health Sciences, University of Science and Technology, Sana'a, Yemen*

[3] *Pediatrics Department, Faculty of Medicine and Health Sciences, Sana'a University, Sana'a, Yemen*

*** Corresponding author: Email:** drahmedhudna@gmail.com

ABSTRACT

Background: Congenital anomalies (CAs) are major causes of morbidity and mortality among newborns worldwide. Therefore, the present study aimed to determine the prevalence and types of CAs in Sana'a city, Yemen.

Methods: This retrospective, cross-sectional study was conducted among newborns admitted to Al-Thawra Modern General Hospital in Sana'a from January to December 2017. Data were collected from 1100 medical records of newborns, besides data about their mothers, using a predesigned data collection sheet. Data were then analyzed, and the results were presented as frequencies and proportions.

Results: Of 1100 newborns, 164 (14.9%; 95% confidence interval: 12.9–17.1) newborns had CAs. Of these, 74.4% had single-system CAs, and 25.6% had multiple-system CAs. Gastrointestinal tract anomalies (43.9%) were the most frequent CAs, followed by anomalies of the central nervous system (CNS) (18.9%), musculoskeletal system (17.1%), chromosomes (15.2%) and cardiovascular system (14%). However, orofacial anomalies of cleft lip and cleft palate (2.4%) were the least frequent CA, followed by urogenital anomalies (8.5%). Esophageal atresia (33.3%) and imperforated anus (25%) were the most frequent gastrointestinal CAs.

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On the other hand, meningocele (35.4%) was the most frequent CNS anomaly. Of musculoskeletal anomalies, limb defects were the most frequent (96.4%), while achondroplasia was observed among 3.6% of newborns with CAs. Congenital heart disease was the most frequent cardiovascular anomaly (95.7%), while dextrocardia was observed among 4.3% of newborns. Hypospadias (57.2%) was the most frequent urogenital anomaly, followed by ureteropelvic junction obstruction (21.5%). More than half of newborns with CAs died, while 35.4% of them were discharged from hospital without correction of CAs. On the other hand, CAs were corrected for 11.6% of newborns before discharge.

Conclusion: The prevalence of CAs among Yemeni newborns is relatively high and can be observed in more than one in ten newborns. The majority of CAs usually affect a single system, most frequently affecting the gastrointestinal tract, followed by the CNS, musculoskeletal system, chromosomes and cardiovascular system. Half of newborns with CAs usually die in hospital before discharge. Therefore, it is necessary to educate parents and conduct regular antenatal screening by a pediatrician to detect these anomalies and manage them appropriately and in a timely manner.

Keywords: Congenital anomaly ▪ Newborn ▪ Yemen

1. Introduction

Congenital anomalies (CAs), also called birth defects or congenital malformations, refer to any defects in morphology, function, biochemistry or molecular structure that may develop in an embryo or fetus from conception until birth.[1] These defects may be inherited genetically, acquired during gestation, or occurred during delivery.[2] CAs can be detected during pregnancy by ultrasonography or laboratory testing or they can be observed at birth or later in life.[3] These anomalies can cause morbidity, disability and mortality for affected children.[4] However, morbidity and mortality associated with CAs can be reduced by detecting and treating these defects in the neonate period.[5] Around 2-3% of newborns and 20% of stillborn fetuses can have major CAs.[6] Globally, about 240,000 infants die within four weeks of birth every year due to CAs.[7,8]

CAs can be an isolated abnormality or part of a syndrome.[9,10] Different classifications of CAs have been used internationally, including single vs. multiple system CAs and major *vs.* minor CAs.[11] Major CAs are those defects that significantly impair normal body functions or reduce life expectancy if uncorrected or uncorrectable, while minor CAs are those that cause no significant physical or functional effect and can be considered as normal variants if affecting over 4% of the population.[12] Annually, 9 million infants (approximately 7% of all births worldwide) are estimated to be born with a serious CA.[13] Moreover, about three million fetuses and infants are born each year with major CAs.[14] Globally, approximately 6% of newborns each year can have a CA of genetic origin. As a result, approximately three million neonates die before they reach their fifth year of age, and other three million are disabled.[14]

The prevalence of CAs varies from country to country, being 1.1% in Japan, 1.5% in South Africa, 1.6% in Lebanon, 2–3% in the United States, and 4.3% in Taiwan.[15] Congenital heart defects, neural tube defects (NTDs) and Down syndrome are the most common CAs.[16,17] It is estimated that 50% of CAs are of unknown origin, and the etiology is genetic in 30–40% and environmental in 5–10% of cases.[3] Genetic causes include chromosomal abnormalities

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(6%), single gene disorders (25%), and multifactorial (20–30%).[3] Environmental factors, such as drugs, radiation, viruses and smoking, can contribute to the incidence of CAs. In Yemen, CAs represent a major problem due to their high social and medical burden. To the best of our knowledge, no studies have been published on the prevalence and types of CAs in the country. Therefore, the present study aimed to assess the prevalence and types of CAs in newborns at a tertiary care hospital in Sana'a city, the capital of Yemen.

2. Methods

2.1. Study design, population and setting

This retrospective, cross-sectional study was conducted among newborns admitted to Al-Thawra Modern General Hospital in Sana'a from January to December 2017. A total of 1100 medical records of newborns were investigated.

2.2. Data collection

Data about the gender of the newborns, the presence and type of CA and its outcome, as well as maternal age, residence, literacy status, parity and consanguinity, were collected using a pre-designed data collection sheet.

2.3. Data analysis

Data were analyzed using IBM SPSS Statistics, Version 20 (IBM Corp., Armonk, NY, USA), and the results were presented as frequencies and proportions.

3. Results

3.1. Prevalence of congenital anomalies

Of 1100 newborns admitted to Al-Thawra Modern General Hospital, 164 (14.9%; 95% confidence interval: 12.9–17.1) newborns had CAs. Of these, 74.4% had single-system CAs, and 25.6% had multiple-system CAs.

3.2. Characteristics of newborns with congenital anomalies

Table (1) shows that the majority of newborns with CAs were males (64%) and urban residents (59.1%). On the other hand, the majority of newborns' mothers were aged 30 years and younger (74.4%), illiterate (52.4%), multigravida (55.5%) and part of a consanguineous marriage (57.3%).

Table 1: Characteristics of newborns with CAs at Al-Thawra Modern General Hospital, Sana'a city, Yemen (2017)*

Characteristics		*n* (%)
Gender		
	Female	59 (**36.0**)
	Male	105 (**64.0**)
Residence		
	Rural	67 (**40.9**)
	Urban	97 (**59.1**)
Mother's age (years)		
	≤30	122 (**74.4**)
	>30	42 (**25.6**)
Maternal literacy status		
	Illiterate	86 (**52.4**)
	Literate	78 (**47.6**)
Maternal parity		
	Primigravida	73 (**44.5**)
	Multigravida	91 (**55.5**)
Parental consanguinity		
	Yes	94 (**57.3**)
	No	70 (**42.7**)

* The total number of newborns was 164; CAs, congenital anomalies.

3.3. Types of congenital anomalies by system

Table (2) shows that gastrointestinal tract anomalies (43.9%) were the most frequent CAs, followed by those in the central nervous system (CNS) (18.9%), musculoskeletal system (17.1%), chromosomes (15.2%) and cardiovascular system (14%). However, cleft lip and cleft palate orofacial anomalies (2.4%) were the least frequent CA, followed by urogenital anomalies (8.5%).

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Table (2) also shows that esophageal atresia (33.3%) and imperforated anus (25%) were the most frequent gastrointestinal CAs. On the other hand, meningocele (35.4%) was the most frequent CNS anomaly, followed by hydrocephalus and meningocele or hydrocephalus (29% each). Of musculoskeletal anomalies, limb defects were the most frequent (96.4%), while achondroplasia (3.6%) was the least frequent. Congenital heart disease was the most frequent cardiovascular anomaly (95.7%), while dextrocardia was observed among 4.3% of newborns. Hypospadias (57.2%) was the most frequent urogenital anomaly, followed by ureteropelvic junction obstruction (21.5%).

3.4. Outcomes of congenital anomalies

Table (3) shows that 53% of newborns with CAs died, while 35.4% of them were discharged from hospital without correction of CAs. On the other hand, CAs were corrected for 11.6% of newborns.

4. Discussion

According to this hospital-based study, roughly 15% of newborns had CAs, with approximately two-thirds being males. The predominance of CAs among male newborns in the present study is in line with that documented for newborns in Iran, Uganda and the United States.[18–20] In addition, newborns of mothers aged <30 years had a higher prevalence of CAs. Maternal age plays an important role in the birth of a congenitally malformed fetus.[21] In Turkey, anomalous births were more frequent in older mothers.[21,22]

Meanwhile, multigravida mothers accounted for more than half of newborns with CAs, which is consistent with findings reported elsewhere.[23–25] However, more CAs were observed among Bengali newborns born to primipara mothers.[26] More than half of newborns with CAs in the present study were born to illiterate mothers, and many studies have suggested that the educational level of mothers may be associated with the presence of CAs.[27-29]

Table 2: Types of CAs among newborns admitted to Al-Thawra Modern General Hospital, Sana'a city, Yemen (2017)*

Types of anomalies	*n* (%)
Central nervous system anomalies	
Hydrocephalus and meningocele	9 (29.0)
Hydrocephalus	9 (29.0)
Anencephaly	1 (3.3)
Meningocele	11 (35.4)
Encephalocele	1 (3.4)
Total	**31 (18.9)**
Orofacial anomalies**	
Cleft lip and cleft palate	**4 (2.4)**
Chromosomal anomalies**	**25 (15.2)**
Cardiovascular anomalies	
Congenital heart disease	22 (95.7)
Dextrocardia	1 (4.3)
Total	**23 (14.0)**
Gastrointestinal anomalies	
Omphalocele	2 (2.8)
Esophageal atresia	24 (33.3)
Tracheoesophageal fistula	4 (5.6)
Gastroschisis	7 (9.7)
Diaphragmatic hernia	2 (2.8)
Imperforated anus	18 (25.0)
Hirschsprung disease	2 (2.8)
Jejuna atresia	8 (11.1)
Duodenal atresia	5 (6.9)
Total	**72 (43.9)**
Urogenital anomalies	
Polycystic kidney	1 (7.1)
Ureteropelvic junction obstruction	3 (21.5)
Multicystic dysplastic kidney	1 (7.1)
Hypospadias	8 (57.2)
Ambiguous genitalia	1 (7.1)
Total	**14 (8.5)**
Musculoskeletal anomalies	
Limb defect	27 (96.4)
Achondroplasia	1 (3.6)
Total	**28 (17.1)**

* The total number of newborns was 164; CAs, congenital anomalies.
** Data not available for all cases. CAs, congenital anomalies.

Table 3: Outcomes of CAs at Al-Thawra Modern General Hospital, Sana'a city, Yemen (2017)*

Outcome	*n* (%)
Discharge without correction	58 **(35.4)**
Discharge with correction	19 **(11.6)**
Neonatal death	87 **(53.0)**

* The total number of newborns was 164; CAs, congenital anomalies.

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Educated mothers often have better access to information about prenatal care, healthy lifestyles and potential risk factors for CAs. On the other hand, more than half of newborns with CAs were born to consanguineous parents, which is consistent with findings reported in other Arab countries and Iran.[30–32] This finding may be explained by the homozygous expression of recessive genes inherited from common ancestors.

In the present study, approximately three-quarters of CAs were present in a single system. Multiple defects were observed in children who had syndrome disorders, followed by NTD. This finding is consistent with that reported among Egyptian newborns, where 69% of CAs were isolated while 31% were multiple.[33] In Nigeria, single-system CAs accounted for 87.8% of birth defects.[34] This difference could be attributed to variations in the susceptibility of embryos to causative agents or the resistance of the embryo to toxicant agents during development. The differences in the prevalence of CAs across countries and even within the same country may be attributed to several factors, including differences in study design.

In the present study, the most frequent CAs involved the gastrointestinal system (43.9%), particularly esophageal atresia and imperforated anus. CAs involving the CNS were the second most frequent anomalies (18.9%), particularly meningocele and hydrocephalus, followed by musculoskeletal defects (17.1%), chromosomal anomalies (15.2%) and cardiovascular anomalies (14%). The least frequent CAs were orofacial (2.4%), mainly cleft lips and palate, and urogenital system (8.5%), mainly hypospadias and ureteropelvic junction obstruction. In a southern region of Saudi Arabia, anomalies of the gastrointestinal system, CNS and cardiovascular system were the most frequent CAs.[35] In contrast, NTDs, followed by orofacial anomalies, musculoskeletal defects, cardiovascular defects, chromosomal anomalies and urogenital defects, were the most frequent CAs.[36] This high frequency of NTDs could be due to no or low use of folate during periconception and early pregnancy. In Romania, congenital heart defects (33.1%), followed by respiratory tract defects, were the most frequent.[37] In Kenya, a higher prevalence (33.9%) was also found for musculoskeletal defects, followed by 28.1% for CNS defects.[38] CNS anomalies were the most frequently observed CAs in Tanzania (29.8%), Nigeria (6.9%), Palestine (18.8%) and China (20.1%). These differences in prevalence could be attributed to several factors, including genetic factors and the study designs employed.

In the present study, more than half of newborns with CAs died, and approximately one-third of newborns were discharged from hospital without receiving surgical correction. The high rates of mortality and discharge with uncorrected CAs could be partly attributed to inadequate facilities, overburdened neonatal wards in governmental hospitals and poor prenatal diagnosis during pregnancy. CAs have been shown to be the second most common cause of infant deaths and the leading cause of mortality in the post-neonatal period (0.52 /1,000 live births) in the United Kingdom.[39] In Iran, nearly one in every three prenatally diagnosed fetuses with CAs was legally terminated before 20 weeks of gestation.[40] However, the social and legal acceptability of this form of secondary prevention is not without questions. In Bangladesh, 56% of congenitally anomalous babies were born alive, but 39% were eventually discharged in apparently good condition after receiving medical treatment or surgical correction. In contrast, 80% of newborns with CAs in Europe were born alive, with only 2.5% dying in the first week of life. Prenatal diagnosis and medical termination are currently the mainstay of secondary prevention of CAs. In Canada, increased prenatal

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diagnosis and subsequent pregnancy termination have contributed to a reduction in the overall national prevalence rate of CAs between 1998 and 2013.[41]

This study is limited by the fact that it was conducted retrospectively in a single hospital, so the results may not be generalizable to the broader population of newborns in the country. Another limitation was that the potential risk factors associated with CAs could not be analyzed. This was due to the large number of missing values for these factors and the fact that the data were collected for the diagnosis of CAs and not for the study of associated risk factors. Consequently, the present study could not provide a comprehensive understanding of modifiable risk factors related to maternal health conditions, environmental exposures, or genetic predispositions. Therefore, multicenter studies with a comprehensive analysis of risk factors associated with CAs are recommended.

5. Conclusion

The prevalence of CAs among Yemeni newborns is relatively high and can be observed in more than one in ten newborns. The majority of CAs usually affect a single system, most frequently affecting the gastrointestinal tract, followed by the CNS, musculoskeletal system, chromosomes and cardiovascular system. Half of newborns with CAs usually die in hospital before discharge. Therefore, it is necessary to educate parents and conduct regular antenatal screening by a pediatrician to detect these anomalies and manage them appropriately and in a timely manner. Further large-scale studies are recommended to identify risk factors associated with CAs among Yemeni newborns.

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Ethical approval and consent

Not required, but permission to collect data was obtained from the administration of Al-Thawra Modern General Hospital.

Conflict of Interest

The authors declare no conflict of interest associated with this article.

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