



## Original Article



### The Frequency, Types and Risk Factors of Congenital Anomalies in a Tertiary Neonatal Intensive Care Unit (A hospital based study)

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#### Abstract

**Background:** Congenital anomalies (CA) are common causes of infant's and childhood deaths and disability. **Objectives:** The aim of the study is to determine the frequency, describe the types and risk factors of congenital anomalies among newborns admitted to Neonatal Intensive Care Unit (NICU) of Assiut University Children's Hospital. **Study design:** It is a prospective observational study (analytic cross sectional study) was performed and screening of the newborns admitted at NICU of Assiut University Children's Hospital (for 6 months) during the period from 1<sup>st</sup> December 2017 to 31<sup>st</sup> May 2018. The sample included 346 newborns, 173 cases and 173 controls. We collected data using a record checklist and an interviewing questionnaire. **Results:** There were significant differences between cases and controls concerning gestational age (P=0.001), single or multiple babies (P=0.002), residence (P=0.001), consanguineous marriage (P=0.01) and family history of unfavorable outcome (P=0.001). We also found that the most common type of congenital anomalies was gastrointestinal anomalies 63 cases (36.4%) with tracheoesophageal fistula 17 cases (27%) being the most common GIT anomalies. Then the musculoskeletal anomalies being the second common anomalies 14.5% with diaphragmatic hernia 10 cases being the most common in musculoskeletal anomalies followed by other anomalies (22 multiple and 1 Conjoined Twins) 23 cases (13.3%) followed by cardiovascular anomalies 22 cases (12.7%), followed by CNS anomalies 18 cases (10%). **Conclusions:** The frequency of congenital anomalies was 22.97%. The most common anomalies were gastrointestinal anomalies (GIT), musculoskeletal anomalies, multiple anomalies and cardiovascular system anomalies. The risk factors were consanguineous marriage, positive family history, urban areas, full-term and singleton pregnancies.

**Key words:** Congenital anomalies (CA), Risk factors, frequency, types

## Introduction

Congenital anomalies or birth defects can be classified according to the World Health Organization (WHO) as functional or structural anomalies. They can be detected during intrauterine life, at birth or later in infancy [1]. Congenital anomalies are common causes of infant and childhood mortality and morbidity. The World Health Organization (WHO) declared that about 303,000 newborns die yearly all over the world within 30 days from birth because of congenital anomalies [2]. Most of these anomalies occur in middle and low-income countries [3]. In Egypt, the frequency of congenital anomalies was 20/1000 live births in 2009 [4]. The frequency of CA among newborns in Assuit University Children's Hospital was 2.06% in 2007 [5]. Major congenital anomalies cause more costly health care than other hospitalizations [6]. Congenital anomalies may be inherited or secondary to environmental factor or multifactorial [6]. It was also found that

more than half of spontaneous abortions are because of chromosomal abnormalities [7]. The critical period in the pathogenesis of congenital anomalies is the first 4 weeks of development, the first 2 weeks which is associated with major congenital anomalies and also severe damage may cause death of the conception, after 2 weeks of development the exposure to teratogen is associated with minor congenital anomalies as epicanthic fold, growth restriction or abnormalities in the phenogenesis [8, 9].

The aim of the work: is to determine the frequency, describe the types and risk factors of congenital anomalies among newborns admitted to Neonatal Intensive Care Unit (NICU) in one tertiary care center.

## Methods

**Study design:** This is a prospective analytic cross sectional study was carried out in the neonatal intensive care unit. It is a tertiary care center. The center

provides multi-specialist care and serves as a major referral center in Upper Egypt for hospitals within and outside the city. The study was done during the period of 6 months from December 2017 to May 2018.

**Patients:** All the babies admitted to the hospital with congenital anomalies during this period were included. All stillbirths were excluded from this study. The sample was 346 newborns, 173 cases and 173 controls. For each case, a detailed perinatal, maternal and family history was obtained by interviewing questionnaire. Diagnosis of congenital anomalies was based on clinical evaluation of newborn babies by the neonatologist and other appropriate investigations such as radiography, ultrasonography, echocardiography and chromosomal analysis etc... Classification of anomalies is done by systems according to WHO international classification of diseases (ICD10) [10]. Data was entered into excel data sheet and appropriate statistical analysis was

performed. The frequency was calculated by dividing the number of CA during the 6 months (173) by the total number of admission during this period (753)  $\times 100$  [11].

### **Statistical analysis**

Data were analyzed using (SPSS Statistics for Windows, Version 21.0, NY). Descriptive analysis of the whole sample; where data were expressed as mean values and standard deviations for quantitative variables, and numbers and frequencies for qualitative variables. Bivariate analysis was performed to assess associations between various independent variables and the dependent variable (presence of congenital anomalies), chi-square was used to compare the difference in distribution of frequencies among different groups and if number inside the cell was small we used Fisher's Exact Test. Independent sample T-test was used to compare the difference in means among different groups. A significant p value was

considered when it less than or equal 0.05.

**Ethics approval and consent to participate:** This study was approved by the head of the University Children's Hospital, the head of the NICU and by the Ethical Committee for Scientific Research of the Faculty of Medicine, Assuit University. (The approval ID of the ethics committee IRB no: 17300493)

An informed written consent was obtained from all parents' participants included in the study.

## Results

Our study included 346 newborns, 173 cases and 173 controls, of the cases there were 138 full-term and 35 preterm, 109 were males and 64 were females. There were a significant difference between cases and controls concerning gestational age, single or multiple babies, residence, consanguineous marriage and family history of unfavorable outcome. There is an increased risk of congenital anomalies in full-term babies, single babies, urban

areas, consanguineous marriage and family history of unfavorable outcome (previous congenital anomalies, stillbirth, and previous abortions) as shown in table (1). We found the frequency of congenital anomalies was 22.97% as shown in figure (1). We also found that the most common type of congenital anomalies was gastrointestinal anomalies in 63 cases (36.4%) with tracheoesophageal fistula was present in 17 cases (27%) being the most common among GIT anomalies. Then the musculoskeletal anomalies being the second common anomalies in 14.5% with diaphragmatic hernia was present in 10 cases being the most common in musculoskeletal anomalies followed by other anomalies (22 multiple and 1conjoined twin as shown in (fig.3) 23cases (13.3%) followed by cardiovascular anomalies 22cases (12.7%) with PDA was the most common, followed by CNS 18 cases (10%) anomalies followed by urinary system anomalies 15 cases (9%)

followed by respiratory system anomalies 4 cases followed by chromosomal anomalies 2 cases and eye, ear, face and neck anomalies only one case being the least common as shown in table (2)

## **Discussion**

The frequency of congenital anomalies among studied newborns was 22.97%. This was in agreement with Sankar et al., 2017[12] who found the frequency of congenital anomalies among stillbirth was 15.3% in a study in a tertiary care hospital in Kerala India. This was in agreement with Adeboye et al., 2016[13] who found the frequency of CA was 111/1000 in a study in a tertiary medical center, located in Bida in Niger State. This also was in agreement with Jangra et al., 2014 [14] who found a high frequency of CA about 30.18% in a study in a Pediatric Surgery Department of a Tertiary Care Institute of North India. Although the result of this study was not in accordance with Kurdi et al., 2019

[15] who found that the frequency of congenital anomalies was about 4.12% in tertiary care center in Riyadh, Saudi Arabia.

Bhide and Kar, 2018 [16] found that congenital anomalies pooled a frequency about 1.8% in India, this high frequency rate in this study, can be explained by that the NICU of Assiut University Children's Hospital is a tertiary care center that receive cases from different areas all over Upper Egypt, but actually the rate may be more because cases of congenital anomalies that didn't need admission were excluded from the study. The most common congenital anomalies in this study were GIT anomalies 36.4%. The predominant defect was tracheoesophageal fistula (17) cases followed by musculoskeletal anomalies 14.5%. This is in agreement with Abdou et al., 2019 [17] who found that the GIT anomalies were the most common anomalies in a study done in the Pediatric University Hospital Alexandria Egypt during the period November 2015–May

2016. This is in agreement with Takai et al., 2019 [18] who found that the most common affected system with congenital anomalies was gastrointestinal system in a study in the Department of Obstetrics and Gynecology and the Department of Pediatrics (Special Care Baby Unit), AKTH, Kano India. This is against Oluwafemi and Abiodun, 2019 [19] who found that CNS is the most common affected system by CA in a study, done at a level II Neonatal Intensive Care Unit of the Mother and Child Hospital Akure (MCHA) in Ondo State, South western Nigeria. This is also against Giang et al., 2019 [20] who found that the most common type of CA being congenital heart defects in central Vietnam. This is against Ameen et al., 2018 [21] that reported CNS anomalies were the most common in the maternity teaching hospital, Erbil city, Iraq. This is against Ecwochi et al., 2018 [22] who found that is the most affected system with congenital anomalies was Musculoskeletal system. This is also

against Abou EL-Ella et al., 2018[23] who found that the most common affected system was musculoskeletal system in a study in pediatric genetics clinic in the pediatric department-Menoufia university hospital. It was found that chromosomal anomalies - were found in 2cases- and eye, ear, face and neck anomalies- were present in only one case- being the least common. This is against Abou El-Ella et al., 2018[23] who found that eye, ear, face and neck anomalies were the 2nd most common system affected but these results were in agreement with his finding that chromosomal anomalies were only 7%. This was against Shawky and Sadik, 2011[4] who found that the 2nd most common system affected was chromosomal anomalies and the eye, ear, face and neck anomalies were the seventh system affected. Concerning the associated factors with CA in this study it was found that congenital anomalies were significantly higher among consanguineous marriage (57.3%) than

non-consanguineous. This is in agreement with Abdou et al., 2019 [19] who found that 47% of cases with CA versus 38% of controls have positive consanguinity history. This is in agreement with Sunitha et al., 2016 [24] who found that consanguinity was a risk factor for CA. This is in agreement with kheir and Yassin, 2015 [25] who found consanguineous marriage in 54% of cases with CA. This is against Taye et al., 2018 [26] who found consanguinity in 4.8% of cases of congenital anomalies. This is against Rostazemahdeh et al., 2017 [27] who found (25%) of the newborns with CA were from consanguineous marriages, while (75%) were from non-consanguineous marriages in a study in Shahid-Madani hospital in Northwest of Iran. This study found that positive family history of unfavorable outcome was significantly associated with congenital anomalies. This is in agreement with Ameen et al., 2018 [21] who found that previous congenital anomalies were associated with high risk

of congenital anomalies. This is against Faal et al., 2018 [28] who found that only one affected case was associated with positive family history of congenital anomalies. This study also found that the CA were significantly common with urban than rural area. This may be due to environmental pollution in the urban areas which is more than rural areas that can be a risk factor for CA. This is in agreement with Li et al., 2019 [29] who found CA of the kidney and urinary tract were more common in urban areas than in rural areas. This is in agreement with Mombo et al., 2017 [30] who found that CA has a lower incidence with rural areas than urban areas. This is in agreement with Xia et al., 2015[31] who found the incidence of CA is lower in rural areas than urban areas. This is against Genowska et al., 2016 [32] who found the mortality rates due to CA were significantly higher in rural areas than urban areas. This is against Cui et al., 2016 [33] who found death rates due to CA was more in rural areas than urban

areas. This study also found that CA were significantly higher with full-term newborns than preterm newborns. This can be explained by the higher death rates of preterm babies than full-term babies so there was not enough time for referral and diagnosis of CA in preterm cases. This is in agreement with Padmanabhan et al., 2019 [34] who found 77% of malformed babies were full-term. This is in agreement with Bairoliya, and Fink, 2018 [35] who found that CA represented about 58.1% of causes of full-term mortality in early neonatal period. This is in agreement with Fontoura and Cardoso, 2014 [36] who found higher frequency of CA with full-term babies. This against Faal et al., 2018 [28] who found higher frequency of premature and LBW newborns in cases with CA was present. This is against Mekonen et al., 2016 [37] who found that CA was higher with low birth weight newborn. This study found that there was increased frequency of CA with singleton pregnancies versus twin pregnancies.

This can be explained by the increased survival of singleton babies versus twin babies in our community. This is against Best and Rankin, 2015 [38] who found increased rate of congenital heart disease among twins than singletons. This is against Menasinkai et al., 2013 [39] who found higher incidence of CA among same sex twins than singletons.

Limitations of the study: The study has included mostly the major congenital anomalies who needed hospital admission. It represents the congenital anomalies in one tertiary care center not all the centers in Upper Egypt.

### **Conclusions**

The frequency of congenital anomalies was 22.97%. The most common anomalies were gastrointestinal anomalies (GIT), musculoskeletal anomalies, multiple anomalies and cardiovascular system anomalies. The risk factors were consanguineous marriage, positive family history, urban

areas, full-term and singleton pregnancies.

We have to intensify the importance of antenatal care and the postnatal screening for CA. We have to improve the awareness in our community about the drawbacks of consanguineous marriage and the importance of genetic counseling in families with positive history. We also recommend further studies with longer duration to discover more congenital anomalies and their preventable causes.

#### **List of Abbreviations:**

ASD: Atrial Septal Defect

CA: congenital anomalies

CHPS: Congenital Hypertrophic Pyloric Stenosis

CNS: central nervous system

GIT: Gastrointestinal tract

ICD10: International Classification of Diseases 10

NICU: Neonatal Intensive Care Unit

PDA: Patent Ductus Arteriosus

TGA: Transposition of great arteries

TOF: Tetralogy of Fallot

VSD: Ventricular Septal Defect

WHO: World Health Organization

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#### **Author's contributions**

AS: designing the work, Data acquisition, interpretation of results and writing the manuscript. AE: Statistical analysis of the collected data and revision of the work. All authors have read and approved the submitted manuscript

#### **Conflict of interest**

Authors declare they have no conflict of interest

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**Table (1): Associated maternal and neonatal factors with congenital anomalies**

Associated factors	Item	Cases	%	Controls	%	P value
<b>Gestational age</b>	Full term	138	58.5	97	41.5	0.001*
	Preterm	35	30.9	76	69.1	
<b>Gender</b>	Male	109	47.6	120	52.4	0.3
	Female	64	53.8	53	47.6	
<b>Single baby or multiple</b>	Single baby	163	52.8	145	47.2	0.002*
	2 or more	10	26.3	28	73.7	
<b>Residence</b>	Rural	83	38.6	131	61.4	0.001*
	Urban	90	68.2	42	31.8	
<b>Maternal age (Years)</b>	Less than 35	147	48.8	154	51.2	0.2
	35 or more	26	57.8	19	42.2	
<b>Drugs received§ during pregnancy</b>	Yes	21	63.6	12	36.4	0.09
	No	152	48.4	161	51.6	
<b>Maternal disease§</b>	Yes	49	48.5	52	51.5	0.2
	No	124	50.6	121	49.4	
<b>Parity</b>	Primiparous	41	47.1	46	52.9	0.5
	Multiparous	132	50.8	128	49.2	
<b>Previous abortion</b>	Yes	48	49	50	51.0	0.86
	No	125	50	124	50.0	
<b>Consanguineous marriage</b>	Yes	86	57.3	63	42.7	0.01*
	No	87	44.2	110	55.8	
<b>Family history of unfavorable outcome</b>	Positive	67	52.8	12	47.2	0.001*
	Negative	106	48.4	161	51.6	

\*P<.0.05 is considered statistically significant

§ (common maternal diseases: Diabetes and preeclampsia) (common maternal drug intake: anti-hypertensives, prenatal steroids and Levothyroxine)

**Table (2): Distribution of GIT anomalies in cases group**

<b>System</b>	<b>Type</b>	<b>No.</b>	<b>Frequency n=173</b>
<b>Digestive system</b> (63)	Tracheoesophageal fistula (TOF)	17	9.8
	Duodenal atresia	10	5.8
	Imperforate anus	8	4.62
	Megacolon	8	4.62
	Jejunal atresia	4	2.31
	Ileal atresia	3	1.73
	Malrotation	3	1.73
	Gastric outlet obstruction	2	1.16
	Congenital hypertrophic pyloric stenosis (CHPS)	2	1.16
	Ranula	1	0.58
	Annular pancreas	1	0.58
	Rectal atresia	1	0.58
	Anal stenosis	1	0.58
	Combined	1	0.58
Colonic atresia	1	0.58	

**Table (3): Distribution of Musculoskeletal anomalies in cases group**

System	Type	No.	Frequency n=173
<b>Musculoskeletal system (25)</b>	Achondroplasia	3	1.73
	Arthrogryposis	1	0.58
	Diaphragmatic hernia	10	5.8
	Cloacal exstrophy	1	0.58
	Bilateral hip dislocation	1	0.58
	Gastroschisis	4	2.31
	Exomphalos major	2	1.16
	Exomphalos minor	1	0.58
	Sacroccygeal teratoma	1	0.58
	Prune belly syndrome	1	0.58

**Table (4): Distribution of CVS anomalies in cases group**

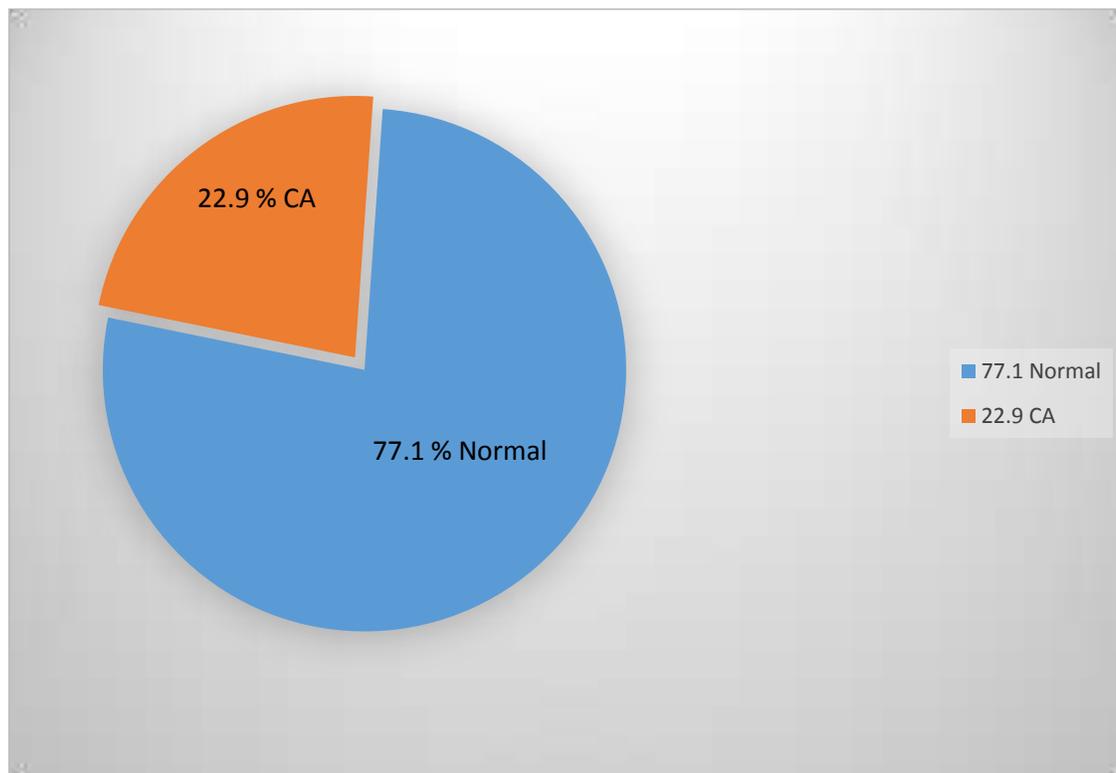
System	Type	No.	Frequency n=173
<b>Cardio-vascular system (22)</b>	Patent Ductus Arteriosus (PDA)	6	3.47
	Atrial Septal Defect (ASD)	5	2.9
	Duct dependent cyanotic heart disease	3	1.73
	Complex	3	1.73
	Ventricular septal defect (VSD)	1	0.58
	Tricuspid atresia	1	0.58
	Extreme fallot's tetralogy	1	0.58
	Total anomalous pulmonary venous return (TAPVR)	1	0.58
	Transposition of great arteries (TGA)	1	0.58

**Table (5): Distribution of CNS anomalies in cases group**

System	Type	No.	Frequency n=173
<b>Central Nervous System (CNS) (18)</b>	Myelomeningocele	6	3.47
	Hydrocephalus	5	2.89
	Combined	1	0.58
	Encephalocele	3	1.73
	Meningocele	3	1.73

**Table (6): Distribution of other anomalies in cases group**

System	Type	No.	Frequency n=173
<b>Other congenital anomalies (23)</b>	Multiple	22	12.72
	Conjoined Twins	1	0.58
<b>Urinary system (15)</b>	Polycystic kidney	5	2.89
	Posterior urethral valve	4	2.3
	Bilateral hydronephrosis	2	1.16
	Multicystic dysplastic kidney disease	2	1.16
	Ectopia vesica	1	0.58
	Combined	1	0.58
<b>Respiratory system (4)</b>	Bilateral choanal atresia	4	2.3
<b>Chromosomal (2)</b>	Down syndrome	2	1.16
<b>Ear, eye, face neck (1)</b>	Microphthalmos	1	0.58



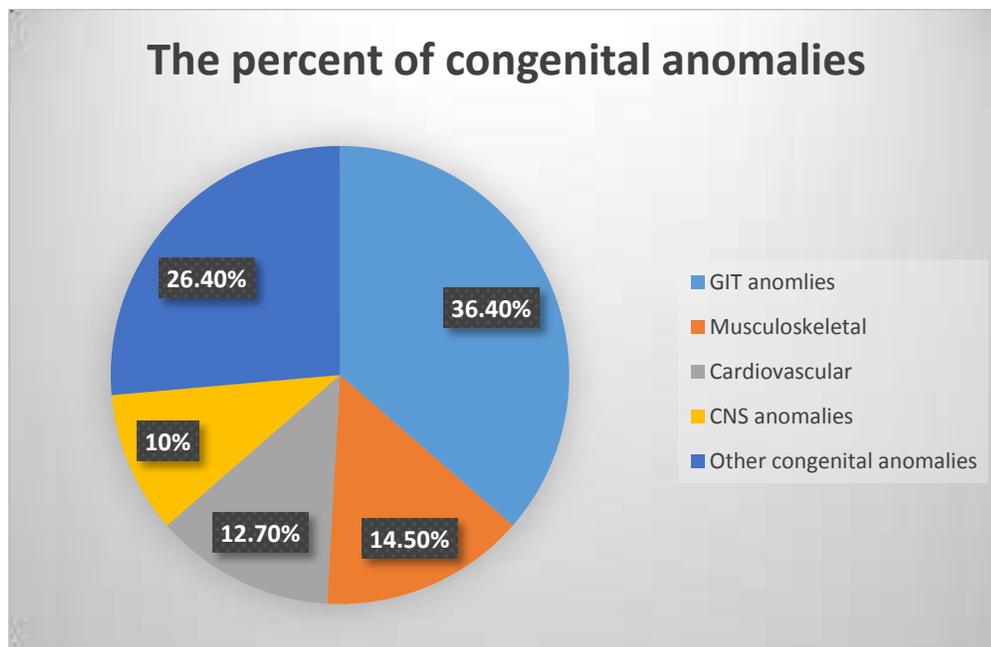
**Fig (1): Frequency of congenital anomalies**



**Fig. (2) Cloacal exstrophy**



**Fig. (3) X-ray of conjoined twins**



**Fig. (4) The Percentage of congenital anomalies**

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