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Research Article

PREVALENCE AND PATTERN OF CONGENITAL ANOMALIES AS SEEN IN A TERTIARY HOSPITAL IN BENIN CITY

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ABSTRACT

Background: Congenital anomalies are a major problem worldwide. They account for a significant cause of morbidity and mortality in low- and middle-income countries. Understanding the burden and pattern of congenital malformation is pivotal in tracking the trajectory and enhancing the healthcare of newborns, particularly in low-income countries. **Objective:** To determine the prevalence and pattern of congenital anomalies seen in babies admitted into the special care baby unit of the University of Benin teaching hospital, Benin City, Edo State, Nigeria. **Method:** This was a hospital-based prospective study carried out on all the neonates admitted into the neonatal unit of the University of Benin Teaching Hospital, Benin City, from January 2018 to December 2021. Data was collected for this study from medical records using a structured questionnaire and analyzed with a statistical package for social sciences version 26.0. **Results:** A total of3755 babies were admitted into the neonatal unit during the study period. Two hundred and eighty-two of them had congenital anomalies (7.5%). Male to female ratio was 1.3:1. Most of the babies were term (82.0%). Among the congenital anomalies observed, those related to the digestive system were the most prevalent (43.0%),followed by anomalies of the central nervous system (23.0%). **Conclusion:** Congenital anomalies continue to pose significant challenges in low- and middle-income countries. Maintaining a high level of suspicion among healthcare professionals is crucial to enable prompt diagnosis and intervention, ensuring timely and effective medical support.

Keywords: Prevalence, congenital, anomalies, Benin.

INTRODUCTION

Congenital anomalies (CA), also known as birth defects, congenital disorders, or congenital abnormalities, encompass structural or functional irregularities that manifest during fetal development.[1] Globally, they contribute significantly to childhood morbidity and mortality, potentially impacting a child's development, survival, and quality of life.[2] Some of these anomalies are readily apparent at birth, while others require the astute diagnosis of attending physicians due to delayed presentation. They encompass a wide range of conditions, including both gross and microscopic malformations, inborn errors of metabolism, mental retardation, and cellular and molecular abnormalities.[3] Congenital anomalies can manifest in isolation as single defects, accounting for approximately 75% of cases.[4] Alternatively, they may manifest as part of recognized associations, such as VACTERL (Vertebral anomalies, Anal atresia, Cardiac defects, Tracheoesophageal fistula, and/or Esophageal atresia, Renal and Radial anomalies, and Limb defects). They can also occur as sequences, characterized by multiple malformations resulting from a single event with diverse underlying causes, as exemplified by the Potter's sequence.[5]

The prevalence of congenital anomalies exhibits regional disparities. In the United States, it is estimated that 2.76% of newborns (or one in every 33 babies) are affected by congenital anomalies each year.[6] In China, this prevalence is higher, with 5.6% of newborns diagnosed with congenital anomalies[7]. Similarly, Canada reported a prevalence of approximately 4% of newborns affected by these conditions.[8] Many developed countries maintain comprehensive birth defect registries that store essential epidemiological data. These registries provide invaluable insights into the types and frequencies of malformations, potential teratogenic environmental factors, and other crucial epidemiological information.

In a systematic review and meta-analysis conducted by Moges *et al*[9] in 2023, the pooled prevalence of congenital anomalies among African studies was found to be 23.5 per 1000 newborns. Anane-Fenin *et al*[10], in a retrospective study conducted at the Special Care Baby Unit of the Cape Coast Teaching Hospital in Ghana in 2023, reported a notably lower prevalence of 2.8%. Meanwhile, in Nigeria, the reported prevalence of congenital anomalies varies, ranging from 2.8% to 15.9%, as indicated in studies conducted by various researchers.[11-17] A more recent study conducted by Chimah *et al*[5] in 2022, among newborns admitted at the Federal Medical Center Asaba, identified a prevalence of 6.1%. It's worth noting that a study within our own hospital, conducted over a decade ago by Okonkwo *et al*[17] in 2011, reported a prevalence of 5.6% among newborns. These varying prevalence rates underscore the need for continued research and surveillance to understand the changing landscape of congenital anomalies in different regions.

CA represent a significant global health concern, contributing substantially to both morbidity and mortality. In 2015, the World Health Organization (WHO)[2] reported that congenital anomalies were responsible for approximately 276,000 newborn deaths worldwide. By 2016, this number had risen to 303,000 neonatal deaths. Annually, about 3.3 million children under the age of five succumb to complications arising from birth defects. Notably, 303,000 of these infants do not survive the neonatal period, and 3.2 million live-born children continue to live with these disabilities throughout their lives. Shockingly, a staggering 95% of deaths globally, occur in low- and middle-income countries.[3-5] This disproportionate mortality in developing nations can be attributed to factors such as inadequate perinatal care, absence of newborn screening tests, exposure to

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teratogens during high-risk periods, or the use of illicit drugs or unconventional medications.[3-5]

Various factors contribute to the occurrence of congenital anomalies, such as maternal age, infections, teratogens, irradiation, consanguinity, or chromosomal abnormalities. However, a significant number of congenital anomalies still have unknown causes, making it challenging to pinpoint the exact etiology.[4] The clinical identification of major congenital anomalies relies on factors like the type of defect, maternal access to healthcare, the method of presentation, and the expertise of the attending healthcare provider.

A notable lack of data exists concerning the occurrence and nature of congenital malformations in our setting. It has now become crucial to determine the prevalence, pattern, and consequences of these malformations in newborns within our local community. The insights gleaned from this research will play a pivotal role in enhancing healthcare planning, subsequently bolstering preventive measures, early detection, and effective management to enhance the overall well-being of these infants. Hence, this study aims to identify the pattern of congenital anomalies seen in the neonatal unit of a major tertiary centre in West Africa, as a follow-up study done in the same setting.

MATERIALS AND METHODS

This study was done in the University of Benin Teaching Hospital, a multispecialty healthcare service provider in Benin City, Nigeria. It is a follow-up of a similar study done at this centre over a decade ago. It's a prospective review of the pattern of congenital anomalies seen in the Special Care Baby Unit (SCBU) of the University of Benin Teaching Hospital, Benin City. The University of Benin Teaching Hospital (UBTH), Benin City is a tertiary health care centre located in the South-South region of Nigeria. It is a centre of referral from within the state and many nearby states of the federation including Ondo, Bayelsa and Delta. The study period was from January 2018 to December 2021. A register of these new-borns with congenital anomalies was kept within the period of the study. A proforma was used to obtain details of the biodata including sex of the neonate, age at the time of diagnosis, maternal age, gestational age of the pregnancy before delivery (term/preterm), baby's birth weight and mode of delivery. The examination findings and the gestational age of the babies were also recorded.The diagnosis of the birth defect was based on the clinical evaluation of newborn babies by the paediatrician as well as information gotten from available Investigations including X-rays, ultrasonography, echocardiography, micturating cystourethrograms, trans-fontanelle ultrasonography, and manometry.

Data processing was done using IBM Statistical Package for the Social Science (SPSS) version 28.The outcomes in the form of morbidity and mortality were also recorded. Babies born at 37 completed weeks were classified as preterm while<37 completed weeks (i.e., <259 days), calculated from the 1st day of the last menstrual period, were considered as premature. Babies born before 28 weeks were considered extreme preterm, those born between 28 and 32 weeks were considered very preterm while those born between 32 and less than 37 weeks' gestation were considered moderate to late preterm. Ethical clearance was obtained from the ethics and research committees of the hospital, and informed consent was obtained from the caregivers.

Data analysis was done using the IBM Statistical Package for the Social Science (SPSS) version. Patients' demography was expressed as percentages using frequency tables and charts. Chi-square was used to test for significance and a*p*-value of < 0.05 was considered statistically significant.

RESULTS

A total of 3755 babies were admitted into SCBU during the study period. The number of babies with congenital anomalies within this period was 282 giving an overall prevalence of 7.5% or 75 babies per 1000 live births.There were 157 males (55%)and124 (45%) females giving a male-to-female ratio of approximately 1.3:1.The demographic features are displayed in Table 1

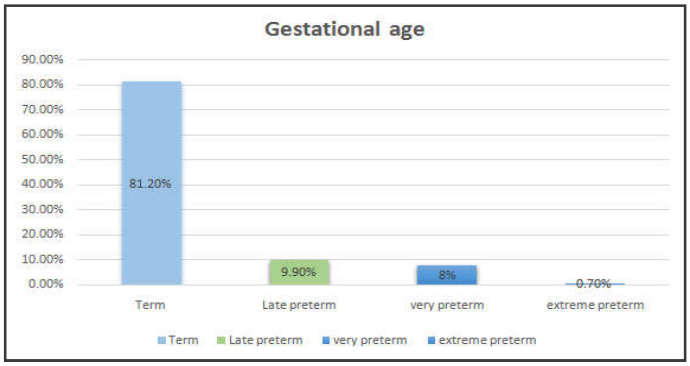
Table 1: Demographic characteristics of the patients

Demographic Characteristics	n	%	X^2	P-value
GENDER				
MALE	157	55.7%	143.809	<0.001*
FEMALE	124	44		
AMBIGOUS GENITALIA	1	0.3		
WEIGHT				
>4200g (high birth weight)	1	0.3	426.789	<0.001*
2500g to 4200g (normal birth weight)	257	91.2		
<2500g (low birth weight)	24	8.5		

n = frequency; % = percent; X^2 = Chi-square Goodness of Fit test; * = significant p value.

Two hundred and twenty-nine of them (81.2%) were term babies while 53 (18.8%) were preterm babies. The majority 28 (52.8%) of the premature babies affected were moderate to late preterm, while 22 (42%) were very preterm. Only 2 (4%) of the extreme pre-terms were found to have congenital anomalies. This is as shown in fig 2 below.

FIG 2: Gestational age



The pattern of the congenital anomalies according to the system involved are shown in table 2 – 8 below.

Table 2: Congenital anomalies by systems

Systems (N = 300)	n	%	X^2	P-value
Central nervous system anomaly	73	25.89	225.560	<0.001*
Digestive system anomaly	122	43.26		
Respiratory system anomaly	23	8.16		
Genitourinary system anomaly	20	7.01		
Musculoskeletal system anomaly	22	7.80		
Cardiovascular system anomaly	13	4.61		
Syndromes	27	10.23		

N = total number; n = frequency; % = percent; X^2 = Chi-square Goodness of Fit test; * = significant p value.

Table 3: Central nervous system anomalies

Anomalies	n	%	X^2	P-value
Myelomeningocoele	45	15.96	194.904	<0.001*
Hydrocephalus	7	2.50		
Spina bifida occulta	1	0.35		
Arnold chiari malformation	8	2.84		
Holoprosencephaly	2	0.70		
Hydrancephaly	2	0.70		
Occipital encephalocoele	4	1.42		
Otocephaly	1	0.35		
Frontonasal encephalocoele	3	1.06		
Total	**73**	**25.89**		

n = frequency; % = percent; X^2 = Chi-square Goodness of Fit test;
* = significant p value.

Table 4: Digestive system anomalies

Anomalies	n	%	X^2	P-value
Omphalocoele	29	10.25	82.098	<0.001*
Gastroschisis	17	6.01		
Duodenal atresia	12	4.27		
Duodenal stenosis	6	2.14		
Annular pancreas	2	0.70		
Anorectal malformation	31	10.95		
Cystic hygroma	2	0.70		
Ileal atresia	11	3.89		
Hirschprung disease	9	3.20		
Congenital bands and adhesions (omental band)	3	1.06		
Total	**122**	**43.26**		

n = frequency; % = percent; X^2 = Chisquare Goodness of Fit test;
* = significant p value.

Table 5: Respiratory system anomalies

ANOMALIES	n	%	X^2	P-value
Tracheoesophageal fistula	19	6.74	40.826	<0.001*
Congenital diaphragmatic hernia	1	0.35		
Laryngotracheomalacia	1	0.35		
Bilateral choanal atresia	2	0.70		
Total	**23**	**8.16**		

n = frequency; % = percent; X^2 = Chi-square Goodness of Fit test;
* = significant p value.

Table 6: Genitourinary system anomalies

ANOMALIES	n	%	X^2	P-value
Posterior urethral valve	11	3.89	23.200	<0.001*
Pelvi-uretheric junction obstruction	2	0.70		
Epispadias	1	0.35		
Bladder extropy	1	0.35		
Hypospadias	4	1.42		
Ambiguous genitalia	1	0.35		
Total	**20**	**7.09**		

n = frequency; % = percent; X^2 = Chi-square Goodness of Fit test;
* = significant p value.

Table 7: Musculoskeletal system anomalies

Anomalies	n	%	X^2	P-value
Port wine stain	1	0.35	8.245	0.201
Congenital ichthyosis	4	1.42		
Congenital talipes equinovarus	6	2.13		
Poly/syndactyly	1	0.35		
Acromelia	1	0.35		
Achondroplasia	4	1.42		
Cleft lip and palate	5	1.77		
Total	**22**	**7.80**		

n = frequency; % = percent; X^2 = Chi-square Goodness of Fit test.

Table 8: Cardiovascular system anomalies

ANOMALIES	n	%	X^2	P-value
Ventricular septal defect	6	2.13	3.923	0.270
Truncus arteriosus	3	1.06		
Atrioventricular septal defect	1	0.35		
Atrial septal defect	3	1.06		
Total	**13**	**4.61**		

n = frequency; % = percent; X^2 = Chi-square Goodness of Fit test.

As seen in tables 2 to 8, the commonest system affected was the digestive system, 122 (43.268%) followed by the nervous system, 73 (25.89%). The least affected systems were the genitourinary and cardiovascular systems, accounting for 7.08 and 4.61% respectively, indicated by P<0.001. Within the digestive system, ano-rectal malformation was the most common condition accounting for 10.99% of all the congenital anomalies, closely followed by omphalocoele (10.25%), indicated by P<0.001. Myelomeningocoele was the single most common congenital anomaly accounting for a total of 45 patients (15.96%).

Table 9: Syndromes

Anomalies	n	%	X^2	P-value
Turners' syndrome	2	0.70	5.345	0.913
Downs syndrome	4	1.42		
Patau syndrome	4	1.42		
Edward syndrome	3	1.06		
VACTERL	3	1.06		
Congenital rubella syndrome	2	0.70		
Eagle Barrette syndrome	3	1.06		
OEIS	3	1.06		
Pierre Robin Syndrome	2	0.70		
Potters sequence	1	0.35		
Beckwith weiderman syndrome	1	0.35		
Goldberg syndrome	1	0.35		
Total	**27**	**10.28**		

n = frequency; % = percent; X^2 = Chi-square Goodness of Fit test.

Table 9 above, shows the frequency of different syndromes seen at our facility during the study period. No statistically significant difference was noted in the prevalence of patients that presented with different syndromes, indicated by P = 913.

Table 10: Outcome

Outcome	n	%	X^2	P-value
DEATH	67	23.76	410.801	<0.001*
DAMA	3	0.70		
DISCHARGE	210	74.50		
ABANDONED	1	0.35		

DAMA = discharge against medical advice; n = frequency; % = percent; X^2 = Chi-square Goodness of Fit test; * = significant p value.

Table 10 above, shows the outcome of babies with congenital anomalies.

Most patients were discharged home, 210 (74.5%), 3 (1.4%) were discharged against medical advice, while 66 (23.8%) of patients died, indicated by P<0.001 The congenital anomalies accounting for the most deaths were gastroschisis, duodenal atresia, and lumbosacral myelomeningocele. To provide a clearer perspective, out of the 17 patients with gastroschisis, 47% did not survive. Similarly, among the 20 patients with duodenal atresia, 40% did not make it, and 17.7% of the 45 patients with lumbosacral myelomeningocele did not survive.

DISCUSSION

Birth defects represent a significant number of health problems within the neonatal period. The landscape of these anomalies within a specific area can evolve over time or by geographical location,which may reflect complex interactions between environmental and genetic issues. It can also beinfluenced by advancements in medical science and the increased availability of advanced screening and diagnostic equipment for identifying congenital anomalies. In this recent study, the prevalence of congenital malformations was found to be 7.1%, a rate higher than the 5.6% reported by Okonkwo *et al*[17] a decade ago, and significantly surpassing figures from various other local studies in Nigeria. For instance, Obu *et al*[11] documented a prevalence of 2.8% among neonates admitted into NBSCU in Enugu, while Ekwunife *et al*[18] reported a prevalence of 2.2% in Nnewi. The elevated prevalence observed in our study, compared to previous studies[11,17,18] can be attributed to improved awareness, enhanced clinical practices, and the utilization of advanced radio-diagnostic evaluations. These advancements have led to the identification of anomalies, especially congenital heart diseases, which might not be apparent during clinical examinations. This highlights the limitation of relying solely on clinical screening, suggesting that it might underestimate the true burden of congenital malformations in our environment. Interestingly, our findings align closely with the prevalence rates reported by Chimah et al[5] (6.1%) and a similar study[12] in southwest Nigeria (6.3%), both of which were also hospital-based studies.

The Prevalence gotten in the present study is however lower than that observed in Maiduguri (13.9%) by JP Ambe *et al*[19] and Wagathu *et al*[20]The significantly lower prevalence noted in this study can be accounted for by better health-seeking behaviours among residents in the southern part of the country, lower rate of poverty, higher rates of female education and also the exposure to better antenatal care services. In this study, the male-to-female ratio was observed to be 1.6:1, a pattern consistent with the previous research conducted in our center (1.7:1). This male preponderance mirrors findings in studies conducted outside the African continent. For instance, a study conducted by Bibi *et al.,* in Pakistan reported a male-to-female ratio of 1.4:1, favoring males. This male preponderance in our study can be attributed to specific congenital anomalies that are predominantly found in males, such as posterior urethral valves, accounting for 3.9% of the congenital anomalies observed in our research. Congenital malformations were found to be more prevalent among term neonates in comparison to preterm infants. This observation could potentially be associated with the increased frequency of full-term deliveries within the study population. It may also be linked to the phenomenon where fetuses with significant congenital anomalies are often lost during the first or early second trimester as spontaneous miscarriages. This discovery aligns with the findings of a study conducted by Chimah *etal*[5] and Takai *et al*[22] but differs from the results reported by Fajolu *et al*[13], who observed a higher prevalence of birth defects among preterm infants.

The pattern of anomalies varies from period to period and from region to region. In our study, anomalies of the gastrointestinal tract had the highest occurrence of 122 (43.26%). The predominance of gastrointestinal anomalies was also documented by several other Nigerian reports.[17-19] This could be attributable to a combination of factors. Firstly, it might be linked to genetic predispositions within the study population that make certain gastrointestinal anomalies more prevalent. Additionally, environmental factors and dietary practices in the region may contribute to this increased occurrence.

A significant majority of infants with anomalies (74.5%) were discharged, indicating that the anomalies observed in our facility were generally non-life-threatening. This outcome suggests the possibility of effective medical and surgical interventions playing a vital role in their treatment and recovery.

CONCLUSION

Congenital anomalies continue to pose significant challenges in low- and middle-income countries. Maintaining a high level of suspicion among healthcare professionals is crucial to enable prompt diagnosis and intervention, ensuring timely and effective medical support.

LIMITATIONS

The lack of a genetic research laboratory and financial constraints among caregivers made it challenging to establish a definitive diagnosis for specific congenital anomalies.

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