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Congenital malformations: Prevalence and characteristics of newborns admitted into Federal Medical Center, Asaba

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Abstract

Background: Congenital malformations account for a significant cause of perinatal mortality and morbidity. Understanding the burden and pattern of congenital malformation is key in monitoring the trend and improving the health care of neonates especially those in low-income countries.

Objective: This was a prospective cross-sectional study to determine the prevalence and characteristics of congenital malformations among neonates admitted to the neonatal unit.

Method: All newborns with congenital malformation admitted into the neonatal unit of Federal Medical Center, Asaba whose parents gave consent were recruited for the study for a 1-year period from January 2020 to December 2020. Appropriately indicated laboratory and radio-diagnostic investigations were done to confirm internal anomalies. Data were collected using a structured questionnaire and analyzed with a statistical package for social sciences version 26.0.

Results: The total admission for the period was 752 with 46 of the neonates (6.1%) having congenital malformation. The predominant system affected was the cardiovascular system (57%), central nervous system (33%), and digestive system (30%). Atrioventricular septal defect (31%) and patent ductus arteriosus (31%) were the commonest types of cardiovascular malformation. A significant number of newborns with congenital anomalies died (43.5%).

Conclusion: Congenital malformation was seen among one in 18 neonates affecting mostly the cardiovascular and central nervous system. A high index of suspicion, early complete physical examination, and radio-diagnostic investigations are relevant for the complete evaluation of CM in neonates. Advanced maternal age was associated with the presence of multiple organ anomalies.

KEYWORDS

birth defects, characteristics, congenital malformation, prevalence

1 | INTRODUCTION

Congenital malformation (CM) or birth defects are alterations in the structure and function of the organ systems of a newborn that occurs in intrauterine life and is identified before, at, or later after birth.[1–3] Birth defects may also be classified as major or minor anomalies depending on the magnitude of the structural and functional disorders and the need for medical support or treatment.[1] Congenital malformation has become a major public health problem because of its significant contribution to newborn and infant mortality and morbidity.[4] The prevalence of congenital malformation varies from one region to another, however global prevalence of CM is about 2%–3%.[5] The prevalence of CM in Nigeria is reported to be between 2.8% and 15.9%,[6–8] which is likely to be underreported as most Nigerian studies on birth defects are mostly retrospective with the limitations of incomplete records.

Etiology of birth defects is documented to be multifactorial and these includes genetic disorders, poor nutrition, TORCHES infection, ingestion of alcohol, and exposure to environmental pollutants such as pesticides, tobacco use, maternal venereal diseases, advanced maternal age, and medical disease conditions in the mothers.[2,9] There is yet no consensus on the definition of advanced maternal age but it is generally accepted that pregnancy in mothers above 35 years is considered high risk.[2,10,11] In addition, single gene defects (6%–7%), chromosomal abnormalities (6%–7%), the interaction between genetics and environmental factors (20%–25%), and exposure to teratogen (6%–7%) have been implicated as the possible underlying cause of birth defects. It is important to note that about 50% of congenital malformations have not been ascribed to any known cause.[2,9]

Congenital malformation affects several organ-system and this depends on the stage of embryogenesis when the insult occurred. Some studies have reported central nervous system anomalies as the most predominant congenital malformation[6,12] while others have reported abnormalities of the digestive system as the commonest congenital malformation.[13,14]

There is a paucity of information on the prevalence and pattern of congenital malformation in our locality. Ascertaining the prevalence, characteristics, and outcome of CM in newborns in our locality has become imperative. Information from this study will help to facilitate better health care planning which will aid in prevention, early diagnosis, and management to improve the quality of life of these newborns. In addition, this will fill the knowledge gap about the prevalence and pattern of CM among newborns in Asaba, Nigeria.

2 | SUBJECTS AND METHODS

The study is a prospective observational study that was carried out in the Neonatal units of Federal Medical Center, Asaba, the capital of Delta State, Nigeria. It was conducted over a 1-year period from January 2020 to December 2020. The hospital provides care for babies born within and outside Asaba, and receives referrals from other parts of Delta State, and from surrounding states including Anambra, Kogi, and Edo states.

All newborns that were delivered in or presented to our facility had a comprehensive physical examination looking out for features of CM within the first 30 min of birth or presentation (for those born outside the facility). The initial examination was done by the admitting pediatric resident and subsequently reviewed and confirmed by the neonatologist in the newborn unit. Anatomical or functional abnormality detected in the organ-system externally or internally was defined as a CM based on WHO definition.[2] Those without obvious physical defects but with clinical suspicion of congenital malformation based on information from the history, clinical presentation, and who required further evaluation were admitted into the newborn unit for laboratory/radiological investigation and management. Specific investigations like computed tomography-scan, abdominal scan, X-ray, and echocardiogram were requested as indicated for each baby. For instance, newborns who presented with cardiovascular symptoms like dyspnea, cyanosis, and murmur were sent for chest X-ray and echocardiography. Only Babies whose parents could afford the relevant investigative test to confirm the diagnosis and who gave consent were recruited for the study. Informed consent was obtained from the parents/guardians of the children before enrollment into the study. A set of standard pretested interviewer-administered questionnaires with close and open-ended questions was used to obtain salient information such as their socio-demographic variables and associated risk factors like an antepartum medical condition. Babies with multiple defects were described as syndromes, sequences or associations based on standard diagnostic criteria.[3] Ethical clearance for the study was obtained from the Ethics committee of Federal Medical Center, Asaba, hospital, the study was voluntary while confidentiality was maintained.

Data were analyzed with Statistical Package for Social Sciences (SPSS) version 26.0. Data presentation was done using frequency tables and figures. The prevalence of CM was determined, maternal features of a newborn with CM was assessed and type of congenital anomaly was also determined while χ^2 and Fisher's exact tests were used to test for significant association of categorical variables were applicable. The level of statistical significance was set at p value <0.05 (Figure 1).

3 | RESULT

A total number of 752 babies were admitted into the neonatal unit of the hospital over the study period, 46 were found to have congenital malformation given a prevalence of 6.1%. Only subjects whose parents gave consent and did the necessary investigation required for diagnosis were recruited.

Table 1 shows the demographic characteristics of mothers whose babies had congenital malformation. Fourteen (30%) belong to the advanced maternal age (more than 35 years), the minimum maternal age was 17 years while the maximum maternal age was

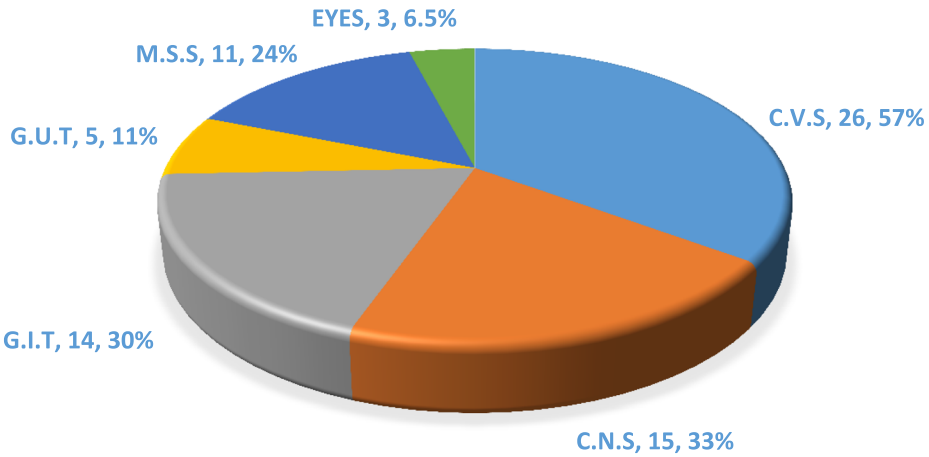

FIGURE 1 Distribution of congenital anomaly: CVS, cardiovascular system; CNS, central nervous system; Eyes; GIT, gastrointestinal system; GUT, genitourinary system; MSS, musculoskeletal system

TABLE 1 General characteristics of mother whose babies had congenital malformation

Variables	Frequency	Percentage
Age (years)		
<19	1	2.2
20–24	5	10.9
25–29	16	34.8
30–34	10	21.7
≥35	14	30.4
Socioeconomic class		
Low	14	30.4
Middle	28	60.9
High	4	8.7
Tribe		
Igbo	17	37.0
Kwale	23	50.0
Urhobo	2	4.3
Ika	4	8.7
Residence		
Asaba	22	47.8
Okwe	1	2.2
Okpanam	1	2.2
Ubulu-Okiti	1	2.2
Ibusa	4	8.7
Agbor	4	8.7
Kwale	2	4.3
Awka	1	2.2
Ogwashuku	2	4.3
Isseleuku	2	4.3

40 years with a mean maternal age of 30.7 ± 5.7 years. The majority of the mothers who resided in Asaba (47.8%), were of the Kwale speaking tribe (50%), and belong to the middle socioeconomic class (57%).

Table 2 shows the clinical characteristics of mothers whose babies had a congenital anomaly. A significant number of mothers 82.6% were booked and were complaint with antenatal medications. Most mothers did not have any antepartum medical condition (82.6%), although Hypertension and Asthma were the most predominant medical condition among the mothers in 4.3%, respectively, while diabetes was seen in 2.2% of mothers. Three mothers received antihypertensive medications while two mothers had used Gin (alcohol) at one point in the antepartum period. A mother had previously delivered a neonate with congenital anomaly and two mothers had a history of previous miscarriages. A significant number of mothers (43.5%) had used herbal medication during the antepartum period, mostly in the first (10.9%) and second trimester (30.4%) respectively.

Table 3 shows the characteristics of babies with congenital malformation. A significant number of babies with birth defects were term (63%) and mostly delivered via SVD (56.5%) with a Male to female ratio of 1:1. Sixty-three percent (63%) had isolated congenital malformation while Downs syndrome was the most common syndrome (64%) associated with congenital anomaly. Other syndromes seen were VACTERL and CHARGE. A significant number of newborns with congenital anomalies died (43.5%).

Table 4 shows the type of congenital anomalies, the cardiovascular system was the system most common affected as seen in 26/46 (57%) babies, closely followed by the central nervous system (33%) and digestive system (30%). Patent ductus arteriosus and atrioventricular septal defect (31%) respectively are the commonest cardiovascular congenital malformation.

Table 5 shows the association between maternal demographic characteristics and the type of newborn congenital anomaly. Maternal age greater than 35 years was associated with multiple

TABLE 2 Clinical characteristics of the mothers whose babies had congenital anomaly

Variables	Frequency	Percentage
Antenatal care		
Booked	38	82.6
Unbooked	8	17.4
Antenatal drugs		
Yes	39	84.8
No	7	15.2
Antepartum condition		
None	38	82.6
DM	1	2.2
Hypertension	2	4.3
Peptic ulcer	1	2.2
Asthma	2	4.3
Eclampsia	1	2.2
HIV	1	2.2
Non-ANC drugs		
Antihypertensive	3	6.0
Gin/alcohol	2	4.3
None	41	89.1
Previous congenital anomaly		
Yes	1	2.2
No	45	97.8
Previous miscarriages		
Yes	2	4.3
No	44	95.7
Herbal drugs use		
Yes	20	43.5
No	26	56.5
Duration of herbal drug use		
First trimester	5	10.9
Second trimester	14	30.4
Third trimester	1	2.2
None	26	56.5

Abbreviation: ANC, antenatal care.

type congenital anomaly, however maternal tribe, socioeconomic status, place of residence, and antepartum use of herbal medication was not associated with the type of congenital anomaly.

Table 6 shows associations between maternal-newborn clinical parameters and the presence of syndromic congenital anomaly. There was no significant association between antenatal booking parameters, compliance to antenatal medication, use of other non-antenatal medications, newborn maturity, and outcome with the presence of syndromic congenital anomaly.

TABLE 3 General characteristics of babies with congenital malformation

Variables		Frequency	Percentage
Gender	Male	20	43.5
	Female	26	56.5
Maturity	Preterm	17	37.0
	Term	29	63.0
Mode of delivery	C/S	20	43.5
	SVD	26	56.5
Outcome	Discharged	22	47.3
	DAMA	4	8.7
	Died	20	43.5
Distribution of CM	Isolated	29	63.0
	Multiple	17	37.0
Associated syndrome	Downs	7	64.0
	VACTERL	3	27.0
	CHARGE	1	9.0

Abbreviations: CHARGE; CM, congenital anomaly; C/S, cesarean section; DAMA, discharge against medical advice; VACTERL: Vertebra, anorectal, cardiac, trachea esophageal, renal anomaly, limb anomaly; VSD, vertex delivery.

4 | DISCUSSION

The prevalence of congenital malformation of 6.1% obtained in this study is high compared to several other local studies in Nigeria. Obu et al.[6] in Enugu documented a prevalence of 2.8% among neonates admitted into NBSCU, while Ekwunife et al.[14] in Nnewi document a prevalence of congenital malformation to be 2.2%. The higher prevalence in our study compared to studies by Obu et al.[6] in Enugu and Ekwinife et al.[14] in Nnewi are apparently due to the utilization of advanced radio diagnostic evaluation of the subjects. This led to more discoveries of internal anomalies especially congenital heart diseases which were not apparent on clinical examination. This shows that clinical screening alone may not be sufficient in detecting some congenital malformation, and may thus underestimate the true burden in our environment.[8] The prevalence of 6.1% in this study is similar to the prevalence rate of 6.3% reported in southwest Nigeria, which was also a hospital based but retrospective research.[7]

Over 80% of the mothers in this study were registered for antenatal clinic and were compliant with antenatal drugs which are prescribed in accordance with the antenatal guideline for favorable pregnancy outcomes which include daily 5 mg of folic acid, 200 mg of fesolate and antimalaria.[11,15] This is not surprising as most of the populace in Asaba belongs to the middle socioeconomic class and has

TABLE 4 Type of congenital malformation seen among the babies

Congenital anomaly	Frequency	Percentage (%)
Cardiovascular system		
Atrioventricular septal defect	8	31.0
Patent ductus arterisous	8	31.0
Atria septal defect	4	15.5
Ventricular septal defect	2	7.8
Tetralogy of Fallot	2	7.8
Dextrocardia	1	3.9
Hypoplastic RT ventricule	1	3.9
Total	**26**	**100**
Central nervous system		
Microcephaly	8	53.3
Meningomyelocele	4	26.7
Encephalocele	2	13.4
Hydrocephalus	1	6.7
Total	**15**	**100**
Gastrointestinal system		
Omphalocele	6	42.9
Gastroscisis	5	35.7
Intestinal atresia	2	14.3
Jejunal web	1	7.1
Total	**14**	**100**
Genitourinary system		
Posterior urethral valve	3	60
Clitoromegaly	2	40
Total	**5**	**100**
Musculoskeletal system		
Talipes equinovarus	5	45.1
Cleft palate	3	27.3
Diaphragmatic hernia	1	9.0
Phocomelia	2	18.0
Total	**11**	**100**
Eyes		
Congenital cataract	3	100
Total	3	100

TABLE 5 Association between maternal demographic characteristics and type of newborn congenital anomaly

Variable	Isolated	Multiple	X^2	*p* Value
Age group				
Greater than 35	3	11	14.959	0.000*
Less than 35	26	6		
Tribe				
Kwale	14	9	0.093	0.760
Others	15	8		
Residence				
Asaba	14	8	0.006	0.936
Others	15	9		
SES				
Low	8	6	3.539	0.170
Middle	20	8		
High	1	3		
Herbal drug use				
Yes	10	10	2.584	0.108
No	19	7		

Note: χ^2 test done.
Abbreviation: SES, socioeconomic status.
*Suggests significant *p* value.

the advantage of getting free maternal treatment as offered by the Delta state government. However, a significant number of mothers also engaged in the practice of herbal medication use in the antenatal period, probably because there is a traditional belief among the people in this part of the country that herbal concoction strengthens the fetus and prepares the mother for easy labor. This finding is supported by the report from Fakeye et al.[16] that over 65% of Nigerian pregnant women use herbal concoctions for various reasons in the antepartum period. Importantly, ingestion of herbal medication did not influence the type of congenital anomaly seen in this study, as both multiple and isolated anomaly was seen amongst mothers who utilized herbal medication. In this study, the congenital malformation involving only one system was higher than those affecting multiple organ system. Furthermore, mothers over the age of 35 years were more likely to have newborns with multiple congenital anomalies. The reason for this is not apparent however, multiple congenital malformations often occurs due to underlying genetic factors or as a sequence in which the primary anomaly leads to several secondary deformation.[3]

The cardiovascular system was the commonest system affected in this study. This finding is similar to a study by Fajola et al.[8] but at variance with other studies which have reported CNS,[6] digestive system[12–14] as the commonest system affected. The difference in the observation is possibly due to the difference in the underlying etiology of the congenital malformation as related to a predominance of downs syndrome and the timing of the insult in relation to embryogenesis. In addition, the pattern of congenital malformation has been shown to vary from period to period and from one location (region) to another. Although, it is not a surprising that the predominant systems affected in this study are the cardiovascular,

TABLE 6 Association between maternal and newborn clinical parameters and presence of a syndromic congenital anomaly

Variable	Syndromic	Nonsyndromic	X^2	*p* Value
Antenatal care				
Booked	13	25	FE	0.613
Unbooked	2	6		
Antenatal drugs				
Yes	13	26	FE	0.589
No	2	5		
Non-ANC drugs				
Antihypertensive	1	2	FE	0.865
Gin	1	1		
None	13	28		
Maturity				
Preterm	5	12	0.125	0.723
Term	10	19		
Newborn outcome				
Discharged	5	17	FE	0.121
DAMA	3	1		
Died	7	13		

Note: χ^2 done.

Abbreviations: ANC, antenatal care; FE, Fishers exact.

central nervous, and digestive systems, as these are the major organ systems affected by inadequate folic acid supplementation. It is not known to what extent the mothers in our environment benefit from pre and peri-conceptional counseling care and nutritional supplementation, especially peri-conceptional folic acid supplementation. A study done in Atlanta showed that mothers who did not get peri-conceptional folic acid supplementation had about 24% higher risk of having a newborn with cardiovascular malformation.[17] Even though, the mothers in this study may have obtained antenatal care at a point in the antepartum period, some of them still engage in traditional practices that may stand in the way of maintaining adequate folic acid stores such as the use of traditional herbal concoction whose unknown content may be antagonistic to folic acid functions. Among the congenital anomalies affecting the cardiovascular system, atrioventricular septal defect (AVSD) and patent ducts arteriosus (PDA) were the two commonest congenital malformations seen in this study. The reason for this, is unknown, however, it might be explained by the high prevalence of Downs syndrome among the children with a congenital malformation in this study.

Downs syndrome was the commonest syndrome seen among children with congenital malformation. This may be partly explained by the advanced maternal age of the women seen in a significant proportion (30%) of mothers in this study. This finding is in support of earlier study by Mashuda et al that reported a high occurrence of congenital malformation among women of advanced age.[18]

Gender distribution of neonates with congenital malformation in this study shows that the prevalence in both genders was comparable. This implies that congenital malformation might not have a predilection for a particular gender. This finding is corroborated by previous studies.[8]

Congenital malformation was more prevalent among the term neonates compared to preterm. The reasons for this, is not clear, however, it may be that the insult to the organ system was probably at a later time in the third trimester or did not threaten the survival of the fetus. This finding is in agreement with a study by Takai et al.[19] but at variance to Fajolu et al.[8] who reported a higher prevalence of birth defect among preterm babies.

5 | CONCLUSION

Congenital malformation accounts for a significant cause of perinatal mortality and morbidity seen among one in 18 neonates admitted in Asaba affecting mostly the cardiovascular and central nervous system. Advanced maternal age was associated with multiple organ anomalies compared with isolated organ anomaly and outcome was not associated with presence of syndromic features. A high index of suspicion, early complete physical examination, and use of necessary radio-diagnostic investigations such as echocardiography, computed tomography, and sonography are relevant for the complete evaluation of CM in neonates.

5.1 | Study limitation

This was purely a hospital-based study although some babies might have been missed because they were not brought to the hospital and even when they presented to the hospital, may not not visit the neonatology unit but are seen at another specialist unit such as pediatrics surgery, neurosurgery, orthopedic clinics, hence not included. In addition, parents were responsible for the payment of relevant radio-diagnostic investigation influencing the prevalence of CM reported as only subjects who could pay for a confirmatory test were recruited.

6 | RECOMMENDATION

A large multicentre/community study will be key in identifying the true burden and pattern of congenital malformation in our environment. Early newborn screening and genetic studies would have been beneficial to classify syndromic congenital anomalies.

AUTHOR CONTRIBUTIONS

Osaretin Uche Chimah: Conceptualization; resources; supervision. **Kennedy Nnaemeka Emeagui**: conceptualization; data analysis. **Obinna Chinedu Ajaegbu**: Methodology; software; writing—original draft. **Chioma Vivian Anazor**: Data curation; project administration.

Adeniyi Johnson Fagbemi: Data curation; validation; visualization. **Omoadoni Diana Emeagui**: Conceptualization; formal analysis; supervision; visualization; writing—original draft. **Chukwuma Anthony Ossai**: Data curation; project administration.

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CONFLICTS OF INTEREST

The authors declare no conflicts of interest.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available and can be accessed by contacting the corresponding author, O. D. Emeagui.

ETHICS STATEMENT

This study was approved by the Ethics Committee of Federal Medical Center, Asaba, Delta State, Nigeria.

TRANSPARENCY DECLARATION

Dr Omoadoni D. Emeagui and other authors declare that this manuscript is an honest, accurate and transparent account of the study being reported; that no important aspects of this study have been omitted.

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