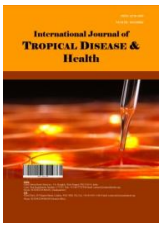

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Prevalence, Pattern and Outcome of Congenital Malformations among Neonates Seen at a Tertiary Health Institution in Yenagoa

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Authors' contributions

This work was carried out in collaboration among all authors. Author IITO principal investigator, conceived and designed the study. Authors OAO and UI participated in writing and technical editing of the manuscript. Authors ASU, KKO, OOA, PPFN and NO collected, contributed and interpreted the data. Authors OEKO and COD contributed in critically reviewing the study and drawing the conclusion. All authors read and approved the final manuscript.

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ABSTRACT

Background: Congenital malformations (CM) are significant causes of childhood morbidity and mortality impacting negatively on the affected family's emotional and financial life. It also results in an enormous burden on a nation's health and socio-economic systems. However, few studies on CM have emanated from developing countries including Nigeria and specifically from Yenagoa, Bayelsa State which is located in the oil rich Niger Delta region. We therefore analyzed the

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prevalence, pattern and outcome of management of babies with CM seen at the Neonatal unit (SCBU) of the Federal Medical Centre, Yenagoa. This could be used as evidence for policy makers to develop and implement CM surveillance, prevention and supportive programs.

Methods: This was a descriptive observational study of all neonates with CM admitted into the Special Care Baby Unit (SCBU) over a one year period from 1st February 2017 to 31st January 2018. Identification and confirmation of congenital anomalies was done by physical examination, diagnostic investigations and surgical interventions. The conditions were classified organ and system-wise except for the chromosomal abnormalities. The prevalence and pattern of defects were determined, while factors related to the outcome of the anomalies were calculated with odds ratio and 95% confidence interval. Data entry and analysis were performed using excel and SPSS version 22.

Results: Among the 502 newborn admissions during the study period, congenital anomalies were found in 61 newborns, giving a prevalence rate of 12.2% with a female preponderance of 55.7%. The commonest CM were those related to the cardiovascular system (47.5%) followed by the digestive system (32.8%) then musculoskeletal system (19.7%). The mean duration of care was 9.7± 9.0 days with mortality of 30.5% recorded at that period. Babies with CM affecting the cardiovascular system and chromosomal anomalies were less likely to survive although these relationships were not statistically significant ($p > 0.05$). However, duration of care showed a statistically significant relationship with outcome as babies who spent one day and less in the neonatal unit had reduced odds of a favourable outcome (OR – 0.07). An increased odd of survival (OR – 2.09) was seen in babies with only one congenital birth defect.

Conclusion: A high prevalence of Congenital malformations in newborns was demonstrated in this study. This has highlighted the need for a well-designed surveillance, prevention and supportive hospital, State and National programme for affected babies and their families.

Keywords: Congenital malformations; neonates; niger delta region; outcome; pattern; prevalence.

1. INTRODUCTION

Congenital malformations (CM) also known as congenital anomalies (CA) or congenital birth defects (CBD), are structural or functional (behavioural and metabolic) defects that occur during intrauterine/fetal life and can be diagnosed prenatally in the fetus, at birth or later in life [1,2]. We considered only structural birth defects of prenatal origin that resulted from defective embryogenesis or an intrinsic abnormality of developmental process in this study. The defects in the morphogenesis of organs could appear as single or multiple. Some of the malformations may also be severe and life threatening, hence contribute significantly to major causes of adverse neonatal outcomes [1-4]. Yenagoa is the capital city of Bayelsa state situated in the oil producing Niger Delta region close to Okolobiri where oil was first discovered in 1956. There are various reports on hazards of exposure to petrochemical products among the residents of oil-rich Niger Delta region due to environmental degradation during the process of crude oil exploration, extraction, refining, transport, and usage [5,6]. These exposures have also been associated with adverse effects on fetal development [5,6].

The true global prevalence of birth defects is unknown due to the lack of national and regional birth defects surveillance systems [1]. However, it has been reported that every year, an estimated 8 million babies (which is about 7% of all births) worldwide are born with serious birth defects and more than 90 percent of these babiesqa3WSZa1q are in low-and middle-income countries [1]. The prevalence of CM differs depending on the period, racial, genetic, geographical and environmental factors [1,7,8]. Hence, while it accounted for 2.4% of 1.5 million annual births in 22 European countries as reported by The European Surveillance of Congenital Anomalies (EUROCAT) and 1 out of every 33 births annually (3% of live births) in USA, the reports emanating from Africa such as Ahuka et al. [9-11]. in Democratic Republic of Congo and Ndibazza et al. [3] from Entebbe, Uganda reported prevalences of 0.41%. and more than 7% respectively. Similar challenges observed with obtaining data on CM worldwide also exist in Nigeria. The data obtained were mainly hospital-based studies (irrespective of the general population) reporting prevalence of 0.4 to 11.1% with no reported peculiarities in the different geographical zones [6,12-16].

The predominant affected systems varied even within the same regions and locality. The central nervous system (0.56%) followed by gastrointestinal tracts (0.37%) predominated as the area most affected in a tertiary hospital in Niger Delta [6]. In a South Western Nigerian study as well as in USA, Lebanon and the United Kingdom [8,17-20] the cardiovascular system was identified as the site for the most common malformations followed by anomalies of the digestive system in other similar studies [8,17-20]. However, some other studies from Nigeria and North-East India reported that the digestive system was the most common system affected [14,21-23]. The skeletal and nervous system have also been identified as predominating in some self reported malformations [3,12,15,24,25]. Considering gender disparity, various studies have reported gender related malformations such as was described by Tennant P et al [26] who reported a male preponderance of congenital anomalies. Congenital malformations account for major causes of child mortality with global average of 9% of under 5 mortality and about half of these deaths in the neonatal period [1]. Global neonatal deaths from congenital anomalies were reported to increase from 260,000 (7%) in 2004 to 303,000 (11.6%) in 2016 probably because of better facilities for diagnostics [27,28]. Meanwhile in Nigeria the reports have been as low as 0.9-1.1% in 2017 [29]. The sequelae for survivors may consist of physical or mental challenges which may constitute emotional and financial difficulties on the affected family and an enormous burden on a nation's health, educational and socio-economic systems [1-4].

It is a fact that considering the enormous challenges posed by congenital birth defects in the low and middle income countries including Nigeria, access and availability of appropriate healthcare services for interventional therapy and rehabilitation cannot be overemphasized. There is a need therefore to determine the prevalence and description of congenital malformations in order to create awareness and plan interventions.

2. METHODS

This was a prospective study of babies presenting with structural congenital anomalies at the neonatal unit of the Federal Medical Centre, Yenagoa. The babies were recruited into the study over a one year period (1st February 2017 to 31st January 2018) and followed up on outcome of the hospitalization. The data was anonymized to maintain confidentiality during analysis.

A structured proforma was used as a guide to obtain information concerning the birth defects, the patients' biodata, history, examination findings and management. Information on prenatal/foetal diagnosis was scanty. Some information on parent's socio-demographic features, full perinatal history could not be obtained due to incomplete information by the caregivers who were often times relatives. Hence, the assessment of risk factors was limited to the responses given. The pattern of the abnormalities was based on physical examination and diagnostic interventions (radiography, ultrasonography, echocardiography and exploratory laparatomy) which was limited to those that were affordable and accessible before death/ discharge. Congenital malformations were classified according to systems adapted from the International Classification of diseases XVII and Bhide et al study [30,31]. The term "unclassified" was utilized for conditions that diagnosis could only be established using clinical examination findings. No laboratory/imaging studies/ surgical interventions could be done on these "unclassified" patients before leaving. A follow-up of the patients during hospitalization was carried out to document their outcome. Data for some of the months could not be obtained because of conditions of industrial action.

Data were analyzed using Statistical Package for Social Sciences version 22.0 (SPSS 22). Frequency distribution tables, means, standard deviations, simple proportions and percentages for different variables were used in the descriptive analyses. The prevalence of CM admissions was determined as the proportion of neonates with CM among the total number of neonatal admissions during the study period. The association between related variables and CM were tested using the Chi-square test. (χ^2) and the *P*-values. The strength of association was estimated using the odds ratio and the corresponding 95% confidence interval. Binary logistic regression analysis was used to determine multiple independent variables that were associated with favorable outcome. The level of significance was set at $p<0.05$ thus when a *P*-value of <0.05 was obtained, the difference between two variables was said to be statistically significant.

3. RESULTS

Sixty one (61) neonates out of a total of 502 admitted neonates had CM resulting in the prevalence of 12.2% in the one year period under review. The females were more in number with 34 (55.7%) and the mean age at presentation was 3.6 ± 4.8 days. The firstborn babies had the highest occurrence of CM with 22 (36.1%) followed by the third born with 12 (19.7%) and then the second born with 11 (18.0%).

The commonest CMs in order of decreasing frequency as shown in Table 1 were related to the cardiovascular system in 29 (47.5%) neonates; digestive system in 21 (34.4%) and musculoskeletal system in 16 (26.2%). The predominant CMs in these systems were ventricular septal defect, omphalocele and polydactyly among 10.3%, 23.8% and 25.0% of the respective systems. In decreasing order of frequency, CM affecting 1, 2, 3 and greater than 3 systems were 67.2%, 18.1%, 9.8% and 4.9% respectively. The difficulties in making specific diagnosis among the different categories of CM, where in 44 (72.1%) of cases.

Table 2 shows that there is a statistically significant relationship between the occurrence of CM and gender (X^2 = 6.37; p – 0.013). When the Odds ratio was calculated CM was almost two times more likely to occur in the female child when compared to the male child (OR – 1.99; 95%CI: 1.12 to 3.55; p – 0.013).

The outcome of care of the neonates with CM in order of decreasing frequency consisted of discharged, died, discharges against medical advice (DAMA) and referred cases in 45.9%, .41.0%, 8.2% and 4.9% of them respectively. There was a statistical significant association of CM with 30.5% of mortality recorded in the new-born unit in the period under review (X^2= 35.23; p – 0.001). The calculated odds ratio of 5.12 (95%CI: 2.72 – 9.12; p – 0.001) implies that children with CM are five times more likely to die when compared to children without CM.

Table 3 explores the factors related to the favorable outcome of care. However, only duration of care shows a statistically significant relationship with favorable outcome of care. The babies who spent one day and less in the neonatal unit had a reduced odd of favourable outcome (OR – 0.07;95%CI:0.01 – 0.38; p – 0.002). Congenital malformations in the cardiovascular system (OR – 0.86) and Chromosomal CM (OR – 0.44) were less likely to be discharged alive, however, this relationship was not statistically significant (p > 0.05). Babies with only one CM showed an increased odd of survival (OR – 2.09), this odd of survival decreases with increasing number of CM present in the child.

4. DISCUSSION

The overall prevalence of CMs in this study was 12.2%. This high prevalence could be attributed to the fact that our institution provides health services to more than 60% of patients and clients in Bayelsa state and nearby communities. The facility is also located in the Niger Delta oil exploration/production region which has had reports on pollution and contamination of water, soil and air with heavy metals such as manganese, arsenic, mercury, cobalt; various gases including carbon monoxide, sulfur dioxide, ozone and nitrogen dioxide; volatile organic compounds (ethylbenzene, benzene, xylene and toluene) [5]. The hazards of exposure to these petrochemical products (environmental teratogens) on fetal development and outcomes of pregnancy among the inhabitants include: maternal or paternal preconception mutagenic action resulting in chromosomal abnormalities and congenital abnormalities [5]. There could also be post-conception teratogenic effects on pregnancy during embryonic or fetal period leading to CA [5,6]. A similar high prevalence of 11.1% and 13.9% were reported by Adeyemo et al in Ibadan, South-West Nigeria and Ambe et al in Maiduguri, North Eastern Nigeria [14,32]. These findings may follow the trend of health conditions in other parts of the world due to better and more efficient diagnostic tools. Another plausible reasons for this similarity could be due to the fact that they were all studies from tertiary hospitals in the same developing country where peculiarities of poor antenatal care, socio-cultural beliefs, harmful religious and traditional practices exist [31–35]. It may also be that the occurrences of consanguineous marriages which are sometimes carried out among the Hausa/Fulanis in Northern Nigeria could lead to easy transmission of inheritable abnormal genes [36]. However, reports from the same South-South geographical region as our centre revealed much lower prevalence of 0.4% by Ekanem et al within Akwa Ibom and Cross Rivers States also 2.07% by Abbey et al in Rivers State [6,15]. These lower prevalences could have resulted from reduced access to hospital based

Table 1. System-wise classification of types of malformations

Affected System	Anomaly	No. of cases	Percent (95% CI of percent)	Prevalence per 1000 neonatal admission
All Anomalies		61	100.0	121.51
Congenital heart defect (CHD)		29	47.5 (33.8 – 57.0)*	57.77
Confirmed CHD	Atrioventricular Septal defect (1), Ventricular Septal defect (2), Cyanotic CHD (1), Patent Ductus Arteriosus (1)	5	8.2 (1.5 – 12.5)*	9.96
Unclassified CHD		24	39.3 (23.3 – 55.3)*	47.81
Chromosomal CBD		10	16.4 (5.9 – 27.9)*	19.92
Confirmed	Down's syndrome	5	8.2 (1.0 – 14.4)	9.96
Unclassified Chromosomal CBD		5	8.2 (1.0 – 16.0	9.96
CM in the Face	Cleft lip (1), Cleft Palate (1), Cleft lip and palate (2)	4	6.6 (1.6 – 13.1)*	7.97
Nervous System Anomalies		8	13.1 (4.3 – 21.5)*	15.94
Confirmed	Hydrocephaly (2), Microcephaly (2), Subarachnoid cyst (1), meningomyelocele (2)	7	11.5 (3.7 – 19.3)*	13.94
Unclassified Nervous System Anomalies		1	1.8 (0.0 – 7.2)	1.99
Genital Anomalies		8	13.1 (4.3 – 21.5)*	15.94
Confirmed	Hydrocele (1), Hypospadias (3), Posterior urethral valve (1), rectovaginal fistula (1)	6		11.95
Unclassified genital anomalies		2	3.3 (0.0 – 8.8)	5.98
Skin Anomalies	Prune belly syndrome (1), Pustular melanosis (1)	2	3.3 (0.0 – 8.8)	3.98
Digestive system CM		20	32.8 (22.3 – 45.5)*	39.84
Confirmed	Duodenal atresia (1), Esophageal atresia (1), Gastroschisis (2), Hirschsprung (2), Hypertrophicpyloric stenosis (1), intestinal obstruction (1), Omphalocele (5), Anorectal malformation (1), pyloric stenosis (1).	15	24.3 (1.0 – 64.5)	29.88
Unclassified Gastrointestinal tract CM		5	8.2 (1.0 – 16.6)	9.96
Eye CM	Unclassified	1	1.6 (0.0 – 4.9)	1.99

Affected System	Anomaly	No. of cases	Percent (95% CI of percent)	Prevalence per 1000 neonatal admission
Auricle CM		3	4.8 (0.0 – 9.6)	5.98
	Absent Auricle	1	1.6 (0.0 – 4.2)	1.99
Unclassified CM in the Auricle		2	3.3 (0.0 – 9.1)	3.98
Musculoskeletal system(MSS) anomalies		12	19.7 (6.6 – 29.5)*	23.90
	congenital absence of one finger (1), constriction bands (1), Over-riding second toe (1) , polydactyl (4), fussed finger (1)	8	13.1 (2.2 – 25.3)*	15.94
	Talipes equino-varus	3	4.8 (0.0 – 10.3)	5.98
Unclassified MSS anomales		5	8.2 (0.0 – 17.9)	
	Unclassified CBD in upper limb (2), unclassified CBD in Lower limb (3)			9.96
Renal anomaly	Hydronephrosis	1	1.6 (0.0 – 6.6)	1.99
Respiratory system anomaly unclassified		1	1.6 (0.0 – 4.2)	1.99
Others		3	8.2 (0.0 – 21.1)	9.96
	Ventral wall hernia (1), Low set ears (1)	2	3.3 (0.0 – 9.1)	3.98
	Unclassified	1	1.6 (0.0 – 6.6)	1.99

**Statistically significant*

Table 2. Association between congenital malformations and sex of children

Characteristics		Congenital malformations		χ^2	df	P-value
	Total N = 502 (%)	Present N = 61 (%)	Absent N = 441 (%)			
Sex						
Male	297 (59.2)	27 (44.3)	270 (61.2)	6.37	1	0.013
Female	205 (40.8)	34 (55.7)	171 (38.8)			

Table 3. Sociodemographic and antenatal factors; CM characteristics; duration of care and their relationship with favorable outcome (discharge alive) among children with CM

Independent variable –	B	OR	95%CI		P-value
(Reference category)			Min	Max	
Age Group (≥ 10 days)					
≤ 1 day	0.87	2.38	0.35	16.36	0.380
2 – 5 days	0.63	1.88	0.25	14.08	0.541
6 – 9 days	1.32	3.75	0.33	42.46	0.286
Sex (Male)					
Female	-0.02	1.02	0.35	2.74	0.973
Locality (Rural)					
Urban	0.38	1.47	0.15	3.03	0.615
CM affecting CVS (No)					
Yes	-0.86	0.42	0.15	1.21	0.107
Chromosomal anomaly (No)					
Yes	-0.44	0.65	0.17	2.52	0.528
Cleft lip and palate (No)					
Yes	0.39	1.48	0.19	11.26	0.706
CM in the Genitalia(No)					
Yes	0.83	2.30	0.42	12.46	0.334
CM in CNS (No)					
Yes	1.35	3.87	0.42	35.36	0.230
CM in the GIT (No)					
Yes	0.06	1.06	0.36	3.16	0.918
CM in MSS (No)					
Yes	1.49	4.42	0.88	22.32	0.072
Number of CM (≥ 4CBD)					
1 CM	0.74	2.09	0.48	9.02	0.324
2 CM	0.25	1.78	0.14	4.35	0.779
3 CM	0.45	1.57	0.13	18.66	0.723
Duration of care (≥ 10 days)					
≤ 1 day	- 2.70	0.07	0.01	0.38	0.002*
2 – 5 days	0.27	1.31	0.27	6.37	0.736
6 – 9 days	0.68	1.97	0.34	11.57	0.453

care for some CMs except probably the ones where interventions may be needed. Ahuka et al. [11] in Democratic Republic of Congo also reported a lower prevalence of 0.41% though it was an increase over previous incidence in the area because of the ongoing civil conflicts.

Varying prevalence emanated from different population within the same geographical area such as South-West, South-East and North-West region of Nigeria with 1.58–6.9%; 0.42-2.8%.and 0.71%-5.8% respectively signifying other multifactorial causes [12,13,17,18,34,35].

A smaller number (32.8%) of the neonates presented with more than one CM per patient which could constitute syndromes. A similar observation of the CMs occurring mostly singly was also reported in this same Niger Delta region, in Northern Nigeria and Congo [6,11,36]. The commonest congenital birth defect site observed in this study was that of the cardiovascular system similar to studies in South Western Nigerian as well as in USA, Lebanon and the United Kingdom [8,18–21]. This was in contrast to the pattern in tertiary hospitals in the same region of Nigeria which reported a predominance of defects of the central nervous system [15,16]. The reasons for this discrepancy cannot be ascertained except for the fact that the limitation in diagnostic intervention may influence the ability to recognize less obvious CAs. However, other prevalence studies from our region and from United States, Lebanon and the United Kingdom agreed with our study of anomalies of the digestive system/ gastro-intestinal tract coming second [6,8,16,18,20,21]. The anomalies of the musculoskeletal system (26.2%) came third although other authors have reported its predominance [3,12,15,24,25].

The relationship between the occurrence of CBD and gender was statistically significant as CBD

was almost two times more likely to occur in the female child when compared to the male child. This finding was supported by Ajao and Adeoye [17] who reported that the male sex was associated with a 16% reduced risk of having a CA though the association between sex and the occurrence of CAs was not significant.

This is in contrast to several studies in the region and internationally which have reported male association [3,13,20,22,23]. The plausible reason for this may be because some of these studies had broadened to include studies on fetuses and stillbirths.

Maternal parity and birth order have been reported to be associated with CAs [37]. Commonly, nulliparity and first birth order as in our study are associated. Environmental risk factors which may not be looked for and biological differences in a nuliparous uterus are said to account for this higher prevalence [38].

Nigeria's ranking globally is second in number of annual neonatal deaths [39]. CAs are reported to contribute greatly to neonatal morbidity and mortality [1,6]. Our study confirmed this as the CM, were significantly associated with 30.5% of the neonatal mortality at the period being studied added to the demonstration of CMs being five times more likely to die when compared to children without CM. Other similar studies showed a lower mortality rate ranging from 3.1 to 16.9% [11,14,17]. Some of these studies cited were community based and differed from our hospital based one that studied patients whose caregivers probably accessed care because of the severity of the CMs hence the high mortality in our study. Among the CMs, it was observed that CMs affecting the cardiovascular system and Chromosomal anomalies were less likely to be discharged alive probably because of limitation with regards to diagnostic interventions. This may have hindered appropriate therapeutic interventions. It is worthy of note that neonates with diagnoses of chromosomal anomalies had associated multiple CAs and our study showed that the odd of survival decreased with increasing number of birth defects present in the child.

5. CONCLUSION

This study showcased the high prevalence (12.2%) of CMs occurring in our setting with its attendant outcome. However, in view of the identified limitations of the study it is highly recommended that a prospective study of CA be carried out over a longer time span and emphasis placed on proper documentation of sociodemographic characteristics, risk factors, medical conditions, family history and accurate obstetric data.

To further our understanding of CMs in the region, future research on the environmental pollutants in the region should be carried out including monitoring the physiological effects, biochemical levels of the pollutants and their metabolites in the neonate.

Institution of CBD record and surveillance system are urgently needed to assist policy-makers to formulate and implement evidenced - based public health interventions. It is important to identify not only the initiators, damage promoters and modulators of CBD, but also the processes that decrease the risk and promote evidence/science based interventions.

It is highly recommended that a CBD multidisciplinary committee be created in the hospital made up of dedicated obstetricians, neonatologists, lab scientists, radiologists, Genetists, counselor and paediatric surgeons.

6. LIMITATIONS

Haphazard documentation and inability to carry out diagnostic intervention resulted in some missed congenital anomalies.

CONSENT AND ETHICAL APPROVAL

This study was approved before commencement by the Federal Medical Centre, Yenagoa Research and Ethics Committee. Verbal consent was obtained from the caregivers together with the consent that is given by patients presenting at the facility agreeing for their data to be used if necessary for research and training while still maintaining confidentiality.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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