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Congenital Birth Defects, Its Incidence and Management Challenges in a Poor Resource Health Facility: A Scenario from North - Eastern Nigeria

Simon Pius[1, *], Machoko Yenti[1], Samuel Wabada[2], Halima Abubakar Ibrahim[1], Mohammed Abatcha Bukar[1], Abubakar Ali Kullima[3], Bisumang Joel Dada[1], Mustapha Bello[1]

[1]Department of Paediatrics, University of Maiduguri Teaching Hospital, Maiduguri, Nigeria
[2]Department of Surgery, University of Maiduguri Teaching Hospital, Maiduguri, Nigeria
[3]Department of Obstetrics and Gynaecology, University of Maiduguri Teaching Hospital, Maiduguri, Nigeria

Email address

simonpius2000@yahoo.co.uk (S. Pius)

[*]Corresponding author

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Abstract: Birth defects also known as congenital anomalies, congenital malformations or congenital disorders are structural or functional anomalies that occur during intrauterine life. They are important cause for neonatal morbidity and mortality and larger percentage of stillbirth.. A mother who gives birth to an abnormal baby is seen as a reproductive failure and may be saddled with the high cost of taking care of such a child. The aim of the study was to determine the incidence and pattern of congenital malformations in UMTH, identify subgroups that are affected and challenge associated with management, provide information that can be used for prevention and counseling. This cross-sectional descriptive study was carried out at the Special Care Baby Unit (SCBU) of the Department of Paediatrics in association with the labour ward of the Department of Obstetrics and Gynaecology of the University of Maiduguri Teaching Hospital, Maiduguri (UMTH), over 18 month period from 1st January 2017-30th June 2017. Diagnosis of BD was based on clinical evaluation, radiographic examination, and echocardiography and karyotyping, whenever recommended. Those diagnosed with birth defects were admitted into the SCBU for observation and documentation. During the 18 months study period, the total admission into Special Care Baby Unit (SCBU) was 1256 neonates, out of these 115 (9.2%) neonates were diagnosed with congenital birth defects, giving the incidence of 9.15/1000 live birth, the male were 53 and female 67 with ratio of 0.8:1. Majority of the birth defects 59 (51.3%) documented consisted of (gastrointestinal tract) digestive system defects, this was followed by central nervous system (CNS) 34 (29.6%), also urogenital system (UGS) and cardiovascular system 4 (3.5%) each, in addition there were 3 (2.6%) cases of CHARGE syndrome. Multiple congenital birth defects constituted 11 (9.6%) of the cases identified. Thirty two (27.8) died before surgical, and 55 (47.8%) that had surgery, 18 (32.7%) died, and 36 (23.8%) were stillbirth with various defects. However, the overall mortality was 50/115 (43.5%), while 56.5% was discharged.

Keywords: Congenital, Birth Defects, Incidence, Pattern and Outcome, Maiduguri

1. Introduction

Birth defects also known as congenital anomalies, congenital malformations or congenital disorders are structural or functional anomalies that occur during intrauterine life and can be identified prenatally, at birth or later in life [1]. A mother giving birth to a malformed baby poses a great dilemma not only to her immediate family, but also to the entire community. A mother who gives birth to an abnormal baby is seen as a reproductive failure and may be saddled with the high cost of taking care of such a child, and thus, children in developing countries born with severe birth defects could be abandoned, concealed, neglected and even murdered by their own parents [2, 3]. They are important

cause for neonatal morbidity and mortality also has been attributable to larger percentage of stillbirth.

Congenital birth defects are disorders of antenatal origin which may be caused by single gene mutation, multifactorial, chromosomal abnormalities, environmental teratogenic substances and deficiency of micronutrients. Maternal rubella, diabetes mellitus, iodine and folic acid deficiency, certain medicines, substance abuse like alcohol and tobacco, chemicals, and irradiation are other factors leading to congenital anomalies. Congenital anomalies, birth defects and congenital abnormalities are terms used for the defects present at birth [4-6]. The occurrence of congenital anomalies has also been associated with advanced maternal and paternal age, parental consanguinity, increasing birth order and low birth weight. About 60% of the causes of congenital anomalies in humans is still unknown [7, 8]. The frequency and specification of congenital anomalies differ from one country to another and from one area to another, it is also depends on their definition, method of detection, duration for which the population was observed, ethnic and socio-economic circumstances of the population studied [9].

Congenital anomalies are classified as major and minor, major require surgical intervention or even can lead to death of the neonate. Minor congenital anomalies are detrimental to quality of life and effects health of neonate [7, 10]. In the USA, congenital anomalies reportedly affect 2-5% of all live births. The magnitude of congenital anomalies in Asia has been shown to vary with reported incidences of 2.5% in India and 1.3% in China [11]. In the Middle East, where consanguineous marriages are common, the prevalence of major congenital anomalies is reported to be 2–2.5%, the highest prevalence (7%) being found in consanguineous marriages [12]. In Africa, some of the rare studies on congenital anomalies have reported an incidence between 1.5% and 2.5%, and most of the studies have been retrospective hospital-based studies which are usually affected by underreporting and other sources of ascertainment bias [7]. In some parts of Nigeria, available reports have prevalence rates ranging from 0.75%-13.9% [1, 13, 14]. The World Health Organization's recent global disease burden (GBD) study reports that anomalies or birth defects rank 17th in the causes of disease burden [15]. They are among the common cause neonatal emergencies and admission and prolonged stay in the special care baby unit (SCBU) in this part of the world.

The aim of the study was to determine the incidence and pattern of congenital malformations in UMTH, identify subgroups that are affected and challenge associated with management, provide information that can be used for prevention and counselling.

2. Subjects and Methodology

2.1. Study Area

This cross-sectional descriptive study was carried out at the Special Care Baby Unit (SCBU) of the Department of Paediatrics in association with the labour ward of the Department of Obstetrics and Gynaecology of the University of Maiduguri Teaching Hospital, Maiduguri (UMTH), which is a level III health facility in North‑East Nigeria. The SCBU admits newborns as referrals from other hospitals in addition to newborns delivered at the centre and admitted for neonatal birth defects and other various illnesses. The SCBU is run by two consultants, a senior registrar, two registrars, two interns and three nursing staffs on every shift. We conducted a prospective study over 18 month period from 1st January 2016-30th June 2017.

2.2. Study Population

All babies delivered in the hospital during the period of study were included. Also all babies delivered at peripheral hospital with congenital birth defects referred to this health facility were included. All neonates delivered in the labour rooms were examined for birth defects (BDs) soon after birth. Diagnosis of BD was based on clinical evaluation, radiographic examination, and echocardiography and karyotyping, whenever recommended. Those diagnosed with birth defects were admitted into the SCBU for observation and documentation. Data collected included the socio-demographics of the parents, gestational age, sex of the babies; ethnicity, birth order and consanguinity were documented.

2.3. Ethical Consideration

The study was approved by the research and ethics committee of the institution. The study was also conducted in strict compliance with 1945 Helsinki declaration on study involving human subjects.

2.4. Informed Consents

Informed consents either verbal or signed were obtained neonates mother or baby caregiver.

2.5. Data Analysis

The data was recorded in a pre‑designed proforma Data were analysis was by using Statistical Package for Social Sciences (SPSS statistics for Windows version 16.0 Ill Chicago USA). Results for categorical variables were expressed using tables and charts while continuous data were expressed using mean and standard deviations where appropriate. Associations between categorical data were determined using Pearson Chi square test. Statistical significance was inferred at p- value < 0.05. The results were expressed in frequencies, means, percentages, tables, figures and charts. The institution's Research and Ethics Committee approved the study.

3. Results

During the 18 months study period, the total admission into Special Care Baby Unit (SCBU) was 1256 neonates, out of these 115 (9.2%) neonates were diagnosed with congenital

birth defects, giving the incidence of 9.15/1000 live birth, the male were 53 and female 67 with ratio of 0.8:1. The mothers age was (18-43±15.25) years, the age the fathers was (30-62±23) years. There were 15 (13.4%) cases of marriages of first cousins. Fifty seven (49.5%) booked and attended to ANC in the study centre while the remaining 58 (50.5%) were unbooked. The mothers were mostly from peri-urban farming community and some others were urban settlement, however there was no history of exposure to known teratogenic agents like radiation or industrial chemicals. Majority of the birth defects 59 (51.3%) documented consisted of (gastrointestinal tract) digestive system defects, this was followed by central nervous system (CNS) 34 (29.6%), also urogenital system (UGS) and cardiovascular system 4 (3.5%) each, in addition there were 3 (2.6%) cases of CHARGE syndrome. Multiple congenital birth defects constituted 11 (9.6%) of the cases identified see table 1.

Table 1. *Frequency distribution of congenital birth defects over 18 months.*

Serial Number	Types of malformation	Number	Percentage of con. malformation
1.	Gastrointestinal	59	100
	Anterior abdominal wall defect	20	33.9
	Gastric outlet obstruction	21	35.6
	TEF	6	10.2
	Others	12	20.3
2.	Central nervous system	34	100
	Neural tube defect	18	52.9
	Encephalocele	11	32.4
	Hydrocephalus	5	14.7
3.	Urogenital system	04	100
	Posterior urethral valve	1	25.0
	Ambiguous genitalia	2	50.0
	Prune belly syndrome	1	25.0
4.	Cardiovascular system	04	100
	Acyanotic CHD	3	75.0
	Cyanotic CHD	1	25.0
5.	CHARGE Syndrome	03	100
6.	Multiple birth defects	11	100

*CHD=congenital heart disease, CHARGE= Coloboma, Heart defect, Atresia choanae, Renal anormalies, Growth retardation, Ear malformations

Table 2. shows distributions of mortalities among the neonates with congenital birth defects system by system before surgical intervention was sort, 20 (33.9%) were mortality among neonates with gastrointestinal defects, central nervous system birth defects was responsible for 8 (23.5%) mortality, one (25.0%) case of ectopia cordis death and mortality from those neonates with multiple congenital birth defects 3 (27.3%), overall

Table 2. *Mortalities among neonates with congenital birth defects that died before surgery.*

Serial number	Variables	Frequency	Percentages
1.	Gastrointestinal tract	20	100.0
	Gastroschiasis	8	40.0
	Intestinal atresia	5	25.0
	Imperforate anus	4	20.0
	Diaphragmatic hernia	2	10.0
	Trachio-esophagial fistula	1	05.0
2.	Central nervous system	8	100.0
	Ruptured meningomyelocele	4	50.0
	Con. hydrocephalus	2	25.0
	Anancephaly	1	12.5
	Encephalocele	1	12.5
3.	Cardiovascular system	1	100.0
	Ectopia cordis	1	100.0
4.	Multiple congenital defects	3	100.0
	Edward syndrome+VSD	1	33.3
	CHARGES Association	1	33.3
	Patau Syndrome	1	33.3
5	Stillbirths	18	100
	Total	52	100.0

*VSD=ventricular septal defect

Table 3. Shows the distribution of birth defects system by system and who benefited from surgery and associated outcome of the surgical intervention. The details are as follows, 14 cases of omphalocele had repair and out of which 6 (42.9%) died, there were 13 cases of duodenal/jujenal atresia that had surgical repair, 3 (23.1%) also died, 9 cases

of imperforate anus underwent surgery, 4 (44.4%) died. Of the cases of central nervous system, one (50.0%) of the cases of hydrocephalus died, while the only case of anencephaly that was operated up on died shortly after surgery. Also one case of ectopia vesicae died after surgery.

Table 3. *Birth defects, surgical intervention and associated outcome.*

Serial number	Types of malformations	Frequency	Mortality/percentage
1	Digestive system		
	Omphalocele	14	6 (42.9)
	Duodenal/jujenal atresia	13	3 (23.0)
	Imperforate anus with sepsis	9	4 (44.4)
	Gastroschiasis	6	1 (16.7)
	Cleft lip/palate	1	0 (0.0)
	Diaphragmatic hernia	1	1 (100.0)
2	Central nervous system		
	Spina bifida/encephalocele	2	0 (0.0)
	Hydrocephalus	2	1 (50.0)
	Anancephaly	1	1 (100.0)
3	Genitourinary system		
	Hydrocele	1	0 (0.0)
	Ectopiae Vesicae	1	1 (100.0)
	Absent urethral meatus	1	0 (0.0)
4	Multiple congenital malformations	2	0 (0.0)

Table 4 show the various congenital birth defects with the corresponding mortality rates. Gastrointestinal associated mortalities were the highest 9 (50.0%), followed by mortalities due central nervous system birth defects 7 (38.9%), while multiple congenital birth defects were responsible for (11.1%).

Table 4. *Still births with various possible diagnoses of congenital birth defects.*

Serial number	Variables	Frequency	Percentages
1	Gastrointestinal tract	9	(50.0)
	Gastroschiasis	6	(66.7)
	Cleft lip/palate	2	(22.2)
	Complete thoracoabdominal defect	1	(11.1)
2	Central nervous system	7	(38.9)
	Anencephaly	3	(42.9)
	Neural tube defects	3	(42.9)
	Hydrancephaly	1	(14.2)
3	Multiple birth defects	2	(11.1)
	CHARGE syndrome	1	(50.0)
	Down syndrome	1	(50.0)

Out of 115 live neonates admitted with various congenital birth defects, 32 (27.8) died before surgical intervention, and out of the 55 (47.8%) that had surgery, 18 (32.7%) died, and record from the labour revealed that 36 (23.8%) were stillbirth with various defects. However, the overall mortality was 50/115 (43.5%), while 56.5% was discharged.

4. Discussion

According to the world health organization [16], congenital birth defects or congenital malformation contribute significantly to neonatal morbidity and mortality. The incidence of birth defects is 2-3/1000 live births [17]. In this study, the incidence of congenital birth defects was 9.15/1000 live births. This was significantly higher than the earlier reports [17] but also lower than that by Cherian et al [18]. The reason being that their study was 10 years retrospective report with larger patient while this was a one year prospective study and sample size was small. Also some other reported incidence 1.9%, [19] from Indian study revealed findings that were similar to this report. The incidence among stillbirth in this study was 3.6/1000 live births which was in contradiction to the reports by Agopian et al [20] who reported 15.7/%.

In this study, most of the mothers were within the reproductive age group, this was similar to the report by Singh et al [3] majority of the women were within reproductive age. In the case of paternal age range, majority were among young age group and only few were above 50 years. Similar reports were in agreement with this finding [18]. This study reported a consanguineous marriage up to 15% among mothers of neonates with congenital birth defects, similar report has validated this practices among some Northern Nigeria socio-cultural ethnic group [3, 21]. This may one reason among other reasons that contributing to propagation of congenital birth defects. Unlike the practice in developed countries where women of child bearing age take folic acid before getting pregnant, in this society many women of this age group don't access folic acid supplement even during pregnancy [20]. Also in this study barely 50% of the pregnant women booked and attended antenatal care services while the rest had no such an opportunity and that might have contributed to the fact that

most of the diagnosis of birth defects were made at delivery. This is common place in developing country like Nigeria as corroborated by reports by some workers [3, 18] and Ambe et al [22] a decade ago in the same centre.

Patients in this study were children born to mothers from a farming community in the peri-urban settlement and no evidence of exposure to industrial chemicals that are implicated as teratogens. Similar findings has been reported a rural Indian study [19]. It has been reported that use of herbicides, insecticides and rodenticides in the farms to which pregnant women exposed to in the rural farming over time, becomes predisposing factors to development of birth defects. We did not find such causal associations as women in this cultural society do not go to farm, rather stay at home caring for off-springs.

With regard to pattern of congenital birth defects in this study, the most common system involved was gastro-intestinal tract (GIT) 59 (51.3%), followed by central nervous system (CNS) 34 (29.6%), urogenital system (UGS) 4 (3.5%), cardiovascular system (CVS) 4 (3.5%) and multiple birth defects 11 (9.6%) this findings were similar to the report by Ekwenife et al [1]. He reported that gastrointestinal tract as the commonest but in his report, urogenital system was second commonest which was not the case in our report. This finding was contrary to the report by Sarkar et al [23] who reported musculoskeletal system as the highest (33.2%), followed by gastro-intestinal tract as the second highest. The highest mortality recorded was among those with GIT birth defects followed CNS and that from multiple congenital birth defects. Similar statistics were also obtained among those who had surgical repair.

In this study still birth among those with birth defects contributed to 13. 5%. Congenital birth defects have contributed significantly to still birth for long time [19, 20, 24, 25], these report support our findings. It must also be attested that this centre have limited capacity to detect antenatally all forms birth defects probably could have been higher than what we documented. Reporting of babies born with birth defects to hospital also is affected by traditional belief as in this society many children born with birth defects are silenced at home by the father's elders. It is still treated with superstitious belief in this sociocultural society.

5. Conclusion

We conducted a cross-sectional study regarding congenital birth defects over one year at the University of Maiduguri Teaching Hospital. We found the incidence of birth defects the pattern, and the associated mortality both among stillbirth and those who had treatment to be comparable to other studies from the developing countries like Nigeria. It is therefore important to educate the community, health professionals and public health on the need for preconceptional, antenatal screening for risk factors and implement various preventive measures.

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