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# Pattern and outcome of gross congenital malformations at birth amongst newborns admitted to a tertiary hospital in northern Nigeria

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**Abstract** *Background:* Congenital malformation(s) do occur in newborns and are thought to be often responsible for a significant proportion of perinatal morbidity and mortality worldwide.
*Objective:* This prospective study was designed to determine the pattern and outcome of congenital malformation(s) among newborn deliveries admitted to the special care baby unit (SCBU) of the Usmanu Danfodiyo University Teaching Hospital, Sokoto.
*Methods:* All newborns with any form of congenital malformation (s) admitted to the SCBU were recruited for the study for a 2-year period from January, 2011 to December, 2012. For ease of identification and classification, organ systems were used to classify malformations while, the recognized syndromes were classified under others.
*Results:* The total deliveries for the study period were 6,578 while, admission to the SCBU was1165. Twenty four (2.1%, prevalence) of the neonates admitted to SCBU had congenital malformation(s); males were 15(62.5%) and females were 9(37.5%) with male to female ratio of 1.7:1. 6(25%) of the babies were delivered outside while, 18(75%) were delivered in our facility labour room. Most of the observed malformations were seen in the central nervous system and the gastro intestinal tract.
*Conclusion:* There is the need for early diagnosis and treatment to improve the chance of survival for malformed babies. The prevalence of 2.1% in our study is comparable to some previous studies however, a community based/multi-centre studies may illuminate a true prevalence.

**Key words:** congenital malformations, newborns

## Introduction

According to the World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth.[1-3] Congenital malformations are single or multiple defects of the morphogenesis of organs identifiable at birth or at the intrauterine life. The global prevalence[1-3] is about 2-3%. Considering the mechanism for the forms of congenital anomalis,[4] congenital malformations may be classified based on the etiologic, clinical or pathogenetic criteria. There is an intrinsically abnormal developmental process, with the early development of a tissue or organ being arrested, delayed, or misdirected. These anomalies can be caused by a teratogen or abnormalities in a gene, a group of genes, or a chromosome.

Congenital malformations contribute highly to prenatal mortality and postnatal physical defects[5] Aggregates of literature indicates that congenital malformations are present in one of every three babies that die in the world[5] and their effects on a child vary depending greatly on the severity of the defect and whether or not other medical problems are present. The etiology of congenital abnormality may be genetic (30–40%) or environmental (5–10%)[6]. Among genetic causes, chromosomal abnormality makes up about 6%, single gene disorders about 25%, and multifactorial factors 20–30%. In about 50% of cases, the cause is not known[6]. Any insult within the early intrauterine period may result in congenital abnormalities[7]. The prevalence of congenital abnormalities ranges from 1% to over 4% depending on the place and population studied [8]

Congenital abnormality plays a major role in morbidity and mortality of children especially at the perinatal period[2, 4]. The treatment and rehabilitation of these children with congenital abnormality is very costly, hence the need to identify causative and risk factors and prevent them early[2,4], where possible. Early recognition of

anomalies is also important for planning and care. Parents are likely to feel anxious and guilt on learning of the existence of a congenital anomaly and this require sensitive counseling.[2]

To the best of the authors' knowledge, no study on congenital malformation has been carried out in North-Western Nigeria; therefore, there was the need to carry out this study to determine the incidence of congenital malformations and the possible causal associations.

## Subjects and Methods

This prospective study was conducted in the Special Care Baby Unit (SCBU) of Department of Pediatrics at Usmanu Danfodiyo University teaching Hospital, Sokoto: the capital of Sokoto State, Nigeria. This tertiary hospital serves as the referral centre to its three neighboring States and Niger Republic.

The study span a two year period; from January, 2011 to December, 2012. All the newborns that were delivered in our facility were examined for congenital malformations soon after birth. Relevant information regarding maternal age, gestational age, sex, ethnicity, birth weight, birth order and consanguinity was documented. Significant antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications of labor was recorded. Antenatal ultrasonography (USS) findings were noted. Relevant radiological, histo-hematological and genetic tests were carried out. Autopsy was not done on stillbirth and neonatal death due to existing strong aversion for the procedure in the study area. There was no karyotyping due to lack of facility. The author who is also the head of the neonatal unit carried out a thorough general, and where necessary a systemic examination to detect any malformations at birth. Where applicable, ultrasound, 3-D echocardiography, electrocardiogram and chest X-ray amongst others were used routinely to detect congenital anomalies including that of the internal congenital anomalies.

For ease of identification and classification, organ systems were used to classify the malformations while, the recognized syndromes were classified under others. Ethical clearance for the study was obtained from Ethics committee of UDUTH, Sokoto

The results were analyzed as simple percentages, Chi-square test were applicable was used for comparison of data and the level of statistical significance was set at $p<0.05$

## Results

There were 6578 intramural deliveries for the 2-year study period, 107 (1.6%) were still births. One thousand, one hundred and sixty five (1165) babies were admitted to the Special Care Baby Unit during the study period (Table 1); 24 had congenital malformation(s) giving the prevalence of 2.1%. Of the babies admitted with congenital malformations, 15 (62.5%) were males and 9 (37.5%) were females with male to female ratio of 1.7:1. Eighteen (75%) of the babies with congenital malformation were delivered in our facility while, the rest 6 (25%) were delivered outside but, admitted same day. Products of multiple gestations were part of the 1165 babies admitted to the SCBU; 18 sets of twins, 6 sets of triplets and 2 sets of quadruplets (total of 50). Two babies though from different set of twins, had congenital malformation. Table 2 shows distribution and frequency of congenital malformations in relation to various fetal and maternal factors: mean birth weight (kg) of 3.26 ± 0.52 SD with minimum of 1.82kg and maximum of 4.35kg, mean maternal age (years) of 26 ± 6, minimum of 17 years and maximum of 43 years. Table 3 shows maternal birth characteristics and occurrence of congenital malformation: over 60% of the mothers who had babies with congenital malformation were seen (ANC) at third trimester, 79% were delivered vaginally and over 90% had no family history of birth defect. Mothers less than 20 years accounted for 62.5% babies with congenital anomalies while, those between 20 and 30 years had 25% babies with congenital malformation. History of parental consanguinity was present in nine cases of congenital malformations. Babies with congenital anomalies were highest in the second order to third birth order (50%). There was a history of oligohydramnios in 5/24 (20.8%) cases and polyhydramnios in 2/24 (8.3%) cases.

**Table 1:** Congenital malformations: Frequency, sex and birth distribution

| | Total cases | Malformed patients | % |
|---|---|---|---|
| Total admissions to SCBU | 165 | 24 | 2.1 |
| Still births | 137 | 5 | 3.7 |
| Live births | 6441 | 19 | 0.3 |
| Male | 3285 | 15 | 0.5 |
| Female | 3156 | 9 | 0.3 |
| Ambiguous | 1 | 1 | 100 |

**Table 2:** Distribution of Congenital Malformations in relation to various fetal and maternal factors

| Characteristics | No of cases per characteristics | % of total (%) |
|---|---|---|
| *Birth Weight (kg)* | N=24 | |
| <2.5 | 15 | 62.5 |
| 2.6 <4 | 7 | 29.2 |
| >4 | 2 | 8.3 |
| *Gestational age at birth (weeks) N=24* | | |
| <37 | 16 | 66.7 |
| 37<42 | 5 | 20.8 |
| >42 | 3 | 12.5 |
| *Maternal age (years)* | *N=24* | |
| <20 | 9 | 37.5 |
| 21-25 | 1 | 4.2 |
| 26-30 | 2 | 8.3 |
| 31-35 | 2 | 8.3 |
| 36-40 | 6 | 25.0 |
| >40 | 4 | 16.7 |
| *Parity* | N=24 | |
| Primip | 8 | 33.3 |
| Para 1-3 | 5 | 20.8 |
| ≥Para 4 | 11 | 45.8 |

**Table 3:** Maternal birth characteristics and congenital malformation

| Characteristics | N (%) malformation |
|---|---|
| *ANC attendance* | *N=18* |
| First trimester | 4(22.2) |
| Second trimester | 3(16.7) |
| Third trimester | 11(61.1) |
| *Mode of Delivery* | *N= 24* |
| Spontaneous Vertex | 19(79.2) |
| Caesarean Section | 3(12.5) |
| Instrumental | 2(8.3) |
| *Family history of birth defect* | N=24 |
| Yes | 2(8.3) |
| No | 22(91.7) |

Table 4 shows the distribution of congenital malformation according to the organ systems: the highest occurrence was in the central nervous system with 7 cases (29.2%) found, with an incidence of 1.1 per 1000 admissions. Twelve (50%) of the cases were treated and discharged home, 6(25%) died all within 48 hours of admission, 2(8.3%) were referred to the neurosurgical unit of our hospital and 4(16.7%) babies were discharged against medical advice.

**Table 4:** Distribution of congenital malformation

| Category of malformation | n (%) n =24 | Prevalence per 1000 admissions n=6578 |
|---|---|---|
| GIT *Omphalocele* | 2(8.3) | 0.3 |
| *Hirschprung disease* | 2(8.3) | 0.3 |
| CNS *Hydrocephalus* | 4(16.7) | 0.6 |
| *Meningocele* | 3(12.5) | 0.5 |
| CVS *Acyanotic Heart Disease* | 1(4.2) | 0.2 |
| Facial/palatial Anomalis *Cleft lip and palate* | 1(4.2) | 0.2 |
| GUS *Posterior Uretheral valve* | 1(4.2) | 0.2 |
| *Ambigous genitalia* | 1(4.2) | 0.2 |
| Ocular *Cataract* | 1(4.2) | 0.2 |
| Miscellaneous *Nasal polyps* | 1(4.2) | 0.2 |
| *Choanal atresia* | 1(4.2) | 0.2 |
| *Limb deformity* | 1(4.2) | 0.2 |
| *Congenital dislocation of the Hip* | 1(4.2) | 0.2 |
| *Multiple Organ deformity* | 4(16.7) | 0.6 |
| Total | 24(100) | |

## Discussion

There is no previous recorded prevalence of congenital abnormalities in the study area and, by extension; the prevalence of congenital malformation in Nigeria has not been properly documented due to a lack of proper record-keeping. The prevalence of congenital abnormalities of 2.1% observed in this study is similar to the findings of Obu et al[2] and Asindi et al[8], that were 2.8% and 2.2% respectively. International prevalence figures of congenital malformation amongst newborns, though not from population similar to our study are comparable with our finding; studies from India[1] and Iran[9] showed prevalence of 1.91% and 2.46% respectively. The fact that both studies were done in referral institutions where major congenital defects are admitted may offer some explanation for the observed similarities. A study from a regional hospital in Oman[10] gave a prevalence of 1.2%; the study concentrated on minor abnormalities alone which may have accounted for the lower prevalence rate obtained in that study.

Congenital malformation among the stillbirths accounted for 3.7%, which is in agreement with previous reports[1,3] Previous publications have shown that the incidence of congenital anomalies is significantly higher in preterm babies as compared to full term babies and, that there is an increased risk of congenital anomalies in babies with low birth weight[1,11,12]. Our study has shown a male preponderance among congenitally malformed babies and, this was in agreement with previous reports [1,11]. There was a history of consanguineous marriages in 6 cases in our study; facts from literature showed a definite increase in incidence of congenital malformation amongst babies of consanguineous marriages[3,13]. In our study, it was observed that mothers with ages below 20 years and those above 35 years had more babies with congenital malformations however, some previous reports [13] showed no statistically significant association between increased maternal age and congenital malformations. Our results are comparable with previous reports[1,2,13,14], that showed a higher incidence of malformation among the mothers of gravida four and above, this probably indicates that there is linear relationship between incidence of congenital malformation and increase in birth order.

Our data presented, however, might not be the true prevalence of congenital anomalies in the area of study. This is because 75% of the babies with congenital malformation were delivered in our hospital, the rest were referred from other hospitals with poor records. Therefore, the prevalence rate of 2.1% obtained in this study does not reflect the picture in the general population as this was purely a hospital based study. In the study arrear, due to poverty, cultural beliefs and "gate keeping" (i.e. husband's permission is required before going out of the house including seeking for emergency medical care), most pregnant women patronize traditional birth attendants (TBA) who do not keep records of malformed babies. There a strong aversion for autopsy in the study area; the authors are aware that carrying out autopsies on the stillbirths may have increased the number of congenital malformations in other previous reports[15]

In addition, some babies with congenital abnormalities brought to our centre do not present to the neonatology unit but are seen at other specialist units such as paediatric surgery unit or neuro-surgery unit, this might represent the true pattern in other centres in the country. The fate of some of the malformed babies that are born outside the hospital included; visits to traditional healers or left to die at home.

Eighteen mothers out of the 24 that had malformed babies, attended ante-natal clinic (ANC); mothers that were seen only at the third trimester had the highest number of the babies with congenital malformation (61.1%). It is therefore possible that either lack of antenatal care or delay in commencing ante natal-care in the

early period of pregnancy when organogenesis begins, may have contributed to these observed higher numbers at the third trimester.

In our study, central nervous system abnormalities accounted for more of the observed congenital malformations in the babies studied with a prevalence of 1.1 per 1000 admissions, this is in keeping with the other previous reports[15,16]. However, facts from other centres in Nigeria[17,18]. showed more occurrences in the gastro intestinal tract. These differences in the pattern of distribution might be due to paucity in investigative procedures such as karyotyping and aversion for autopsy in the study arrear.

**Conclusion**

The prevalence of congenital malformation in our study was 2.1% with the commonest malformations seen in the central nervous system. The prevalence rate obtained in this study, however, may not reflect the true situation in the general population for reasons given above, therefore, a community based/multi-centre studies may illuminate a true prevalence.
Congenital anomalies are a major cause of stillbirths and infant mortality; there is the need for early diagnosis and treatment for better chance of survival for the malformed babies.

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