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# An 8-year review of major congenital abnormalities in a tertiary hospital in Lagos, Nigeria

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**Abstract***: Background:*
Congenital abnormalities are defects present at birth and are increasingly becoming an important cause of neonatal mortality. They can also result in disability in majority of the survivors.
*Objective:* To describe the pattern and outcome of major congenital abnormalities (MCA) in a tertiary hospital in Lagos, Nigeria.
*Methods:* The labour ward and labour ward theatre delivery records and admission records of the in-born ward of the neonatal unit of a tertiary hospital were reviewed retrospectively from January 2007 to December 2014. The MCA were classified according to the 10th revision of the International Statistical Classification of Diseases and Related Health Problems, (ICD-10). Multiple abnormalities were counted once by the system with the most major anomaly. Abnormalities were grouped according to organ systems, sex and yearly distribution. Statistical analysis was based on systemic type, and neonatal outcome.
*Results:* Out of 14581 deliveries during the study period, 167 had MCA, giving an incidence of 11.5 per 1000 total births; 71 (42.5%) were females, 91 (54.5%) were male and 5(3.0%) had indeterminate sex. There was an increase in the yearly incidence from 2007-2014
The most frequent abnormalities were in the central nervous system (31.7%), musculoskeletal system (18.6%), complex congenital abnormalities group (15.5%) and urogenital system (11.4%). Fifty six (31.8%) infants died in the first week of life contributing 12.6% to the overall early neonatal mortality during the study period. Overall case fatality rate was 42.0%; case fatality was highest in the other abnormalities group, followed by chromosomal, cardiovascular and genitourinary system abnormalities respectively.
*Conclusion:* The incidence of MCA in this study is high especially in the central nervous system. There was a steady increase in the yearly incidence during the study period. The overall case fatality rate was also very high with a high contribution to early neonatal deaths

**Keywords:** Congenital abnormalities; Early neonatal death; Lagos; Nigeria; Teratogens.

## Introduction

Congenital anomalies also known as birth defects, are structural or functional abnormalities, including metabolic disorders, which are present from birth, irrespective of whether the defect is caused by a genetic factor or by prenatal events that are not genetic[1]. These prenatal events can result in an arrest, delay or misdirection in the development of a structure early in embryonic life with a resultant permanent effect. Congenital abnormalities can be minor with minimal or no risk for disability or death, or major with a significant risk of morbidity and mortality. All population share the burden of con-

genital abnormalities although the frequency and type may vary. Globally, congenital disorders are estimated to be present in 7% of all births, with a total of 9 million babies born annually with major congenital abnormalities which can result in death or lifelong disability.[2]

Between 2003 and 2007, the European Surveillance for Congenital Anomalies (EUROCAT) in 22 countries reported a incidence of 23.9 per 1000 births with 80% of births being live births.[3] In Nigeria, the reported incidence of congenital abnormalities vary from 2.1 to5.1% amongst live births in Northern Nigeria[4,5] to 15.9% of all births (including still births) in South west Nigeria.[6]

Congenital abnormalities are a major cause of infant morbidity and mortality and they were reported to have contributed 276,000 deaths (4·4%) in the neonatal period globally in 2013.[7] They may also result in long term disabilities which will ultimately have a significant impact on the child, family, health care system and the society at large.

These abnormalities may be caused by environmental factors such as irradiation, smoking and alcohol consumption in the mother,[8,9] or are inherited via abnormal genes from the carrier or affected parent. Lack of access to medical care, malnutrition, environmental exposure to smoke, alcohol, irradiation, chemical and drugs are important factors in the occurrence of congenital anomalies. The presence of a major congenital abnormality in a foetus or new-born evokes emotional parental responses that require sensitive counselling, hence early recognition of these anomalies is important in planning care for the baby and support for the family to minimise the risk of abuse.

With increasing industrialization, worsening poverty, self-medication practices, patronage of traditional birth attendants and ingestion of herbal concoctions by pregnant women in developing countries like Nigeria; pregnant women are more likely not to take routine antenatal drugs like folic acid which has been shown to prevent abnormalities like neural tube defects. They are also more likely to be exposed to infections like syphilis that increase the risk of congenital abnormalities.[2] There is therefore a need for continuous update on the types of MCA in our environment. This is required in order to follow the trend and variability so as to aid policy decisions especially in the area of prevention and provision of care and support for the affected children and their families and also help in counselling affected families. While a considerable reduction in cases of congenital abnormalities have been achieved in the developed countries; more than 90% of births and 95% of deaths of children with congenital abnormalities occur in developing countries.[2] The aim of this study therefore is to describe the pattern and outcome of major congenital abnormalities in babies delivered at a tertiary hospital in Lagos.

## Methodology

The labour ward and theatre records of all deliveries and the admissions records of all neonates with major congenital abnormalities admitted into the inborn ward of the neonatal unit of the Lagos University Teaching Hospital from January 2007 to December 2014 were reviewed retrospectively. The hospital has a radiology department that is equipped with facilities for X-ray, ultrasound scan, computerized tomographic scan and magnetic resonance imaging. The department of paediatrics in the hospital has an echocardiogram machine with neonatal probes for diagnosing structural abnormalities of the heart. The hospital has facilities for neurosurgical, paediatric surgical and urological surgeries.

All babies delivered in the hospital are examined in the labour ward or in the postnatal ward by a paediatric resident doctor; those with abnormalities or with clinical features suggestive of abnormalities are identified and admitted in the neonatal unit for investigation and management. Other babies with other conditions requiring further care are also admitted. The diagnosis of congenital abnormality was made by clinical examination and / or imaging studies (x-ray, ultrasound, magnetic resonance imaging or computerized tomographic scan) as indicated. Major congenital abnormalities were defined as abnormalities that are life threatening or has the potential of causing disability.
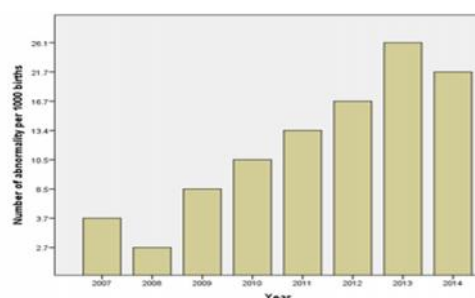
The major congenital abnormalities were then classified by systems according to the 10th Revision of the International Statistical Classification of Diseases and Related Health Problems, (ICD-10);[10] multiple congenital abnormalities were counted only once by the system with the most major anomaly. The congenital abnormalities were further grouped according to organ and systems and by sex and analyzed on yearly distribution. The abnormalities were analysed on the basis of anatomical and systemic type, and also on the basis of neonatal outcome.

Data were analysed using the statistical package of social sciences (SPSS) version 22; frequencies and percentages were generated; the student t-test and chi square were used as tests of statistical significance and a p value of $<0.05$ was considered as significant.

Ethical approval was obtained from the Health research and ethics committee of the Lagos University Teaching Hospital.

## Results

During the eight year review period, there were 14581 deliveries and 167 of these infants had major congenital abnormalities giving an incidence of 11.5 per 1000 total births. There were 71 (42.5%) female, 91 (54.5%) male and 5(3.0%) with indeterminate sex. Fifty-seven (34.1%) of these neonates were born before 37 completed weeks of gestation.

The yearly incidence of congenital abnormalities is shown in Figure 1; the incidence of congenital abnormalities increased over the past eight years of review with the 2014 incidence-(2.17/1000 live births) being greater than five times the 0.37/1000 live births seen in 2007.

**Fig 1:** Trend in yearly congenital abnormality rates

Seventy of the newborns with congenital abnormalities died, giving a case fatality rate of 42.0%. Fifty six of these deaths were early neonatal deaths and they accounted for 12.6% of the overall 446 early neonatal deaths during the study period.

All the cases requiring surgical intervention were co-managed with the surgeons; some were managed conservatively, some had surgeries, some either died before surgery or had to be discharged to allow parents source for funds for surgery later as there are currently no free services for children in the hospital.

The ICD-10 classification of the abnormalities is shown in Table 1; the systems with the most frequent abnormalities were the central nervous (31.7%), musculoskeletal (18.0%) and the urogenital systems (11.4%) and digestive (9.6%) systems respectively. Complex abnormalities that did not fall in any system (classified as other abnormalities by ICD-10 system) also contributed 15.5% to the total number of cases seen.

Table 2 shows the case fatality rates and the percentages of deaths that were early neonatal deaths in the various groups of abnormalities. The case fatality rates were highest in the other abnormalities group, followed by the digestive system, chromosomal abnormality and cardiovascular system, while all the deaths in the respiratory and cardiovascular systems were early neonatal deaths.

**Table 1:** ICD-10 classification of congenital abnormalities seen in Lagos University Teaching Hospital.

| ICD 10 Classification | No | Percentage |
|---|---|---|
| **Q00-Q07-CNS** | 53 | 31.7 |
| Dysraphism (7 Spina bifida, 3 encephalocoele) | 10 | |
| Hydrocephalus (5 with spina bifida) | 39 | |
| Anencephaly | 3 | |
| Poroencephaly | 1 | |
| **Q20-28-Respiratory system** | 5 | 3.0 |
| Pulmonary hypoplasia | 1 | |
| Choanal atresia | 3 | |
| Laryngomalacia | 1 | |
| **Q30-34- Cardiovascular system** | 7 | 4.2 |
| Dextrocardia | 1 | |
| Acyanotic congenital disease | 1 | |
| Cyanotic congenital cardiac disease | 3 | |
| Venous abnormalities | 2 | |
| **Q35-Q45 - Digestive system** | 16 | 9.6 |
| Cleft lip and cleft palate | 5 | |
| Intestinal atresia | 6 | |
| Imperforate anus | 5 | |
| **Q50-Q64-Urogenital system** | 19 | 11.4 |
| Ambiguous genitalia | 5 | |
| Posterior urethral valve | 4 | |
| Multicystic kidneys | 3 | |
| Hypospadias | 3 | |
| Hydronephrosis | 2 | |
| Undescended testes | 2 | |
| **Q65-Q79-Musculoskeletal system** | 30 | 18.0 |
| Skeletal dysplasias | 3 | |
| Limb abnormalities | 10 | |
| Abdominal wall defects | 12 | |
| Prune belly syndrome | 3 | |
| Craniosynostosis | 2 | |
| **Q80-Q89-Other anomalies (Conjoined twins, complex anomalies )** | 26 | 15.5 |
| Q90-Q99-Chromosomal | 11 | 6.6 |
| Trisomy 21 | 5 | |
| Edward's syndrome | 3 | |
| Others | 3 | |

**Table 2:** Case fatality rates in major congenital abnormalities distributed according to specific systems/groups

| System | No of cases (N=167) | No of deaths (N=70) | Early neonatal deaths N (%) | Case fatality Rate (%) |
|---|---|---|---|---|
| Central nervous | 53 | 13 | 9 (69.3) | 24.5 |
| Cardiovascular | 7 | 3 | 3 (100) | 42.9 |
| Respiratory | 5 | 1 | 1 (100) | 20.0 |
| Digestive | 16 | 9 | 6 (66.7) | 56.3 |
| Urogenital | 19 | 6 | 4 (80.8) | 31.6 |
| Musculoskeletal | 30 | 12 | 10 (83.3) | 40.0 |
| Chromosomal | 11 | 5 | 4 (80.0) | 45.5 |
| Others | 26 | 21 | 19 (90.5) | 80.8 |

## Discussion

The overall incidence of congenital abnormality during the study period was 11.5 per 1000 total births. This is lower than the 15.8 per1000 total births reported by Iroha et al in a study[5] in Lagos, Nigeria 20 years ago and the 24 per 1000 births reported in Lebanon[11] in 2009. The slightly higher rate in the earlier study in Lagos may be due to the fact that it included results of autopsy of neonates that died while this present study did not. The incidence from Lebanon rates was quite high and may be explained by the significant number of consanguinity reported amongst parents of infants with MCA and also the significant number of mothers who consumed alcohol amongst the MCA group.

The incidence of MCA in this study was higher than the rates reported from studies from Northern and Southern Nigeria done over a decade ago with an incidence of 5.5/1000 births[5] and 4.0/1000 births[12] respectively. The explanation for this difference is not very clear as consanguinity which has been associated with congenital abnormalities is common in Northern Nigeria and the Southern part of Nigeria where the other study[12] was done is an oil producing area with a higher risk of environmental pollution; thus a higher incidence would have been expected in these areas. Both studies were however done over a decade ago. A study from Iran done about the same period with the current study (2004-2012) however reported a similar incidence (11.2/1000 births).[13]

There was an increase in the yearly incidence during the study period with the incidence for the year 2014 being over 5 times that of 2007. The reason for this may be the problems of urbanisation such as exposure to environmental pollution and modernisation including exposure to radiation from electronic gadget such as mobile phones and computers with its attendant effects on the populace. The association of environmental pollutants such as heavy metals, pesticides and hydrocarbons with congenital abnormalities have been documented.[14] It is therefore expected that the incidence in more industrialised countries should be high; however low rates are reported. This could be misleading considering the fact that facilities for prenatal diagnosis are available with the option of termination of any pregnancy with the diagnosis of congenital abnormality. This is exemplified

in the EUROCAT study where 53% and 33% of spina bifida and Downs syndrome were prenatally diagnosed and terminated.[15]

In Nigeria, many women do not attend antenatal care clinics and when they do, they register late usually after the first trimester when organogenesis would have been completed; thus they miss out on early commencement of folic acid which is known to protect against neural tube defects which had the highest occurrence in this study. Neural tube defects were also the most common abnormality in some earlier studies.[4,12]

The system with the highest frequency of abnormalities in some other Nigerian studies was the gastrointestinal.[5,16] This may be due to difference in the classification of disorders; the current study used the ICD-10 system which is recommended for uniformity in disease classification while other studies did not state the method used in the classification. One of the studies[16] classified omphalocoele under gastrointestinal abnormalities while ICD-10 classifies it under the musculoskeletal system. This is of importance if a congenital abnormality database is to be created so that entries made are the same for specific abnormalities using standard classification systems.

The overall case fatality for congenital abnormalities was 42%, however when the different systems were analysed, the case fatality was highest in the other abnormalities group, most of the babies in this group had complex congenital abnormalities that were not compatible with life and 90.5% of these deaths were early neonatal deaths. All the deaths in the cardiovascular and respiratory systems were early neonatal deaths and occurred before any surgical intervention could be planned. Eighty percent of the deaths among the neonates with MCA occurred in the first week of life, contributing significantly to the early neonatal deaths during the study period; thus interventions to reduce the incidence of congenital abnormalities will go a long way in reducing overall under-five mortality.

Most developed countries have tackled sepsis; have improved services for care of the preterm infant and better access to both antenatal and neonatal care services; hence congenital abnormalities now rank high as cause of infant mortality. Even though these factors mentioned above are more important contributors of mortality in developing countries like Nigeria, it appears that the incidence of congenital abnormalities is rising. Thus reducing the incidence of MCA will go a long way to reduce the infant mortality rate and also reduce the number of affected families who have to cope with either the loss of a child or bear the burden and stigma of caring for a disabled child. In addition, this will also reduce the burden on the community and nation as a whole. A good starting point for the above will be commencement of surveillance for congenital abnormalities in all countries so as to have a database of the pattern and possible aetiology of these congenital abnormalities.

## Conclusion

The incidence of congenital abnormalities in Lagos University Teaching Hospital has increased over the past eight years with a greater than 5-fold increase from 2007 to 2014; the commonest system affected was the central nervous system. Congenital abnormalities also contributed significantly to perinatal mortality.

It is therefore important to educate the populace on simple preventive measures such as folic acid use even before conception and avoidance of possible teratogens such as exposure to radiation (including telephone related radiation), alcohol, certain drugs, tobacco, pesticides and infections like syphilis as these may go a long way in reducing the incidence of congenital abnormalities. There is need for further studies to determine the risk factors, possible causes for this increasing incidence and also the burden of care of these children on the family and community. In addition, a national database for congenital abnormalities needs to be opened and morbidity in these children should also be documented so as to plan and advocate for adequate care for these vulnerable children.

## Acknowledgement

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