Prevalence and Pattern of Birth Defects in the Two Tertiary Hospitals in Enugu, South East Nigeria: A Hospital-Based Observational Study

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Abstract

Background: Birth defects have medical, surgical and aesthetic consequences. The impact of birth defects is particularly severe in low-income countries where health-care resources are limited. **Aims and Objectives:** The aim of this study was to determine the prevalence and types of birth defects in live newborns delivered at the two tertiary hospitals in Enugu, South East Nigeria. **Materials and Methods:** This was a hospital-based observational study carried out on neonates delivered in the maternity units of the University of Nigeria Teaching Hospital Ituku/Ozalla and Enugu State University Teaching Hospital Parklane, Enugu during the periods of January 2015 and December 2018. All the live-born babies born in the two hospitals during this period were recruited into the study. The neonates were examined by a paediatrician for the presence of birth defects. The birth defects were classified according to the system involved and also into major and minor. **Results:** During the study period, 9492 babies were born, of which 166 had birth defects, which gave a prevalence of 1.75%. The predominant system affected by birth defects was the musculoskeletal system (45.2%) followed by the central nervous system (34.9%), urogenital system (10.8%) and gastrointestinal tract (9%). 13 (7.8%) patients had their birth defects diagnosed prenatally during the maternal ultrasound scan. Fifteen (9%) and 13 (7.8%) mothers of the neonates who had birth defects were diabetics and hypertensives, respectively. Most of the neonates were delivered vaginally. **Conclusion:** Birth defects are not uncommon. This study showed a prevalence of 1.75% in the two tertiary hospitals in Enugu, South East Nigeria. The most commonly affected system was the musculoskeletal system.

Keywords: Birth defects, pattern, prevalence, tertiary hospitals

INTRODUCTION

Birth defects, also known as congenital anomalies, congenital disorders, or congenital malformations, are structural anomalies which are present at the time of birth.[1] It has medical, surgical and aesthetic consequences. Birth defects may be classified according to the system involved or categorised into major or minor types. Major birth defects include severe anatomical anomalies that compromise life, while minor birth defects are structural anomalies that have no serious consequences and are corrected by simple surgical techniques.[2] The causes of birth defects include genetic and environmental factors and the interactions between the factors. Environmental risk factors include teratogens such as maternal rubella infection and medications like thalidomide.[3,4] In >50% of the cases, the cause of birth defect is unknown.[5] Birth defects have no geographical boundaries and occur in every country of the world. In the USA, birth defects reportedly affect 2%–5% of all live births and is a leading cause of infant mortality.[6] Birth defects can be a cause of lifelong disability, and the emotional cost to the family cannot be quantified.[7] Little is known about the incidence of birth defects in Enugu, Nigeria. Therefore, this study was undertaken to determine the prevalence and pattern of birth defects in the two tertiary hospitals in Enugu, South East Nigeria.

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MATERIALS AND METHODS

This was a prospective study carried out at the two tertiary hospitals in Enugu, namely: University of Nigeria Teaching Hospital (UNTH) Ituku Ozalla, Enugu and Enugu State University Teaching Hospital (ESUTH), Parklane, Enugu, South East Nigeria. The two tertiary hospitals serve an estimated 5 million people that include Enugu and the surrounding states of Abia, Anambra, Benue, Ebonyi, Delta, Imo and Kogi states. This study covered a period of 3 years, from January 2015 to December 2018. For the purposes of this study, our interest was on the clinically obvious and observable abnormality of structure or form which was present at birth or noticed a few days after birth. All the live babies born in UNTH and ESUTH during the period of this study were included. Stillborns were excluded from this study.

All the neonates had a thorough physical examination (general and systemic) performed by a paediatrician. The diagnosis of birth defect was based only on the clinical evaluation of the newborn babies by the paediatrician. Investigations such as chromosomal analysis, radiography, ultrasonography and echocardiography were not performed. The birth defects were categorised according to the system involved and also into major or minor. For each patient, the following data were collected: sex of the neonate, age at the time of diagnosis, maternal age, gestational age of the pregnancy before delivery (term/preterm), baby's birth weight and mode of delivery. Baby's birth weight greater or equal to 2.5 kg was considered to be normal, while birth weight < 2.5 kg were considered as low birth weight. Babies born at < 37 completed weeks, calculated from the 1st day of the last menstrual period, were considered preterm while babies born at or after 37 completed weeks were considered term. Ethical clearance was obtained from the ethics and research committee of both tertiary hospitals, and informed consent was obtained from all the patients' mothers.

Data analysis

The Statistical Package for the Social Science (SPSS) version 21 (SPSS version manufactured by IBM Cooperation, Chicago, Illinois, USA) was used for data entry and analysis. Data were expressed as percentages and means. Chi-square or Student's *t*-test was used to test for significance. $P < 0.05$ was considered statistically significant.

RESULTS

Patents' demography

During the period of the study, a total of 9,492 neonates were delivered in the two tertiary hospitals in Enugu, among whom 166 neonates had birth defects. 7119 babies were delivered in ESUTH Parklane, Enugu (111babies had birth defects), while 2373 babies were delivered in UNTH Ituku/Ozalla, Enugu (55 babies had birth defects). This gave an overall birth defect prevalence of 1.75% or 17.5 babies per 1000 live births. Other demographic features are shown in Table 1.

System wise distribution of birth defect

The predominant system affected by birth defect was the musculoskeletal system 75 (45.2%) followed by the central nervous system 58 (34.9%), urogenital system 18 (10.8%) and gastrointestinal tract 15 (9.1%). Specific birth defects are shown in Table 2.

Classification of the defects into major or minor

The specific birth defects may also be categorised into major or minor, as shown in Table 3. It is important to note that the division between major and minor is far from perfect. Minor defects can be related to major defects or be indications of certain syndromes.

Prenatal diagnosis of birth defect

A total of 95 mothers (57.2%) whose children had birth defects underwent antenatal ultrasound scan, but only 13 (7.8%) patients had their birth defects diagnosed prenatally during the maternal ultrasound scan. Birth defects were missed during ultrasound scan in 82 (49.4%) patients. Some of the mothers went for the ultrasound scan on their own. Eight (9.5%) out of the 84 mothers that had antenatal care were sent for antenatal anomaly scan. Out of this 8, only 5 (62.5%) were positive for birth defects, while 3 (37.5%) were negative for birth defects. The remaining 8 birth defects (13 minus 5) were discovered as incidental findings during the routine antenatal ultrasound. Gastrointestinal (omphalocele) and central nervous systems (myelomeningocele) anomalies were the most commonly diagnosed birth defects, prenatally.

Booked/unbooked mothers

Among the mothers whose babies had birth defects, 84 (50.6%) were booked, whereas 82 (49.4%) were unbooked. Comparing booked mothers with unbooked mothers gave a value of $P = 0.22$, which is not statistically significant.

Table 1: Demographic profile of the patients	
Parameters	**Number (%)**
Gender (%)	
Male	96 (57.8)
Female	69 (41.6)
Ambiguous genitalia	1 (0.6)
Mean postnatal age at diagnosis	7 days
Maturity at birth (%)	
Term	67 (40.4)
Preterm	99 (59.6)
Weight at birth (kg) (%)	
<2.5	79 (47.6)
2.5- 4	38 (22.9)
>4	49 (29.5)
Maternal age (years) (%)	
<20	7 (4.2)
20- 35	118 (71.1)
36 and above	41 (24.7)
Mode of delivery (%)	
Vaginal	141 (84.9)
Caesarian section	25 (15.1)

Table 2: System wise distribution of birth defects (*n*=166)

System	*n* (%)
Musculoskeletal system	75 (45.2)
Polydactyly	24 (14.5)
Syndactyly	21 (12.7)
CTEV	20 (12.0)
Chest wall deformity	9 (5.4)
Phocomelia	1 (0.6)
Central nervous system	58 (34.9)
Myelomeningocele	29 (17.5)
Hydrocephalus	16 (9.6)
Encephalocele	4 (2.4)
Anencephaly	1 (0.6)
Microcephaly	8 (4.8)
Urogenital system	18 (10.8)
Hypospadias	11 (6.6)
Micropenis	3 (1.8)
Ambiguous genitalia	1 (0.6)
Epispadias	1 (0.6)
Bladder exstrophy	2 (1.2)
Gastrointestinal system	15 (9.0)
Omphalocel	5 (3.0)
Cleft lip/palate	4 (2.4)
Imperforate anus	2 (1.2)
Gastroschisis	2 (1.2)
Intestinal atresia	1 (0.6)
Oesophageal atresia	1 (0.6)

CTEV: Congenital talipes equinovarus

Table 3: Classification of the specific birth defects into major or minor (*n*=166)

Major		Minor	
Category	*n* (%)	Category	*n* (%)
Myelomeningocele	29 (17.5)	Micropenis	3 (1.8)
Hydrocephalus	16 (9.6)	Polydactyly	24 (14.5)
Encephalocele	4 (2.4)	Syndactyly	21 (12.7)
Anencephaly	1 (0.6)	CTEV	20 (12.0)
Microcephaly	8 (4.8)	Chest wall deformity	9 (5.4)
Omphalocele	5 (3.0)	Phocomelia	1 (0.6)
Cleft lip/palate	4 (2.4)		
Imperforate anus	2 (1.2)		
Gastroschisis	2 (1.2)		
Intestinal atresia	1 (0.6)		
Oesophageal atresia	1 (0.6)		
Hypospadias	11 (6.6)		
Ambiguous genitalia	1 (0.6)		
Epispadias	1 (0.6)		
Bladder exstrophy	2 (1.2)		

CTEV: Congenital talipes equinovarus

Associated maternal illness, infections, behavioural and environmental factors

Inquiries showed no evidence of the ingestion of any teratogenic agents such as herbal concoctions during early pregnancy or just before getting pregnant. However, 15 (9%) and 13 (7.8%) mothers were diabetics and hypertensives, respectively. History of maternal infections such as toxoplasmosis, rubella, cytomegalovirus and herpes was not present. Nutritional assessment of the mothers was not done. None of the mothers accepted to be smokers or alcoholics. Undue exposure to environmental agents such as lead, mercury or drug abuse could not be established.

DISCUSSION

Birth defects are inborn errors of foetal development, and it represents a public health challenge because of the impact on population health.[8] The prevalence and pattern of birth defects may vary over time or with geographical location, reflecting a complex interaction of known and unknown genetic and environmental factors.[9] Birth defects reduce the average life expectancy and quality of life of newborns.[10]

The difference in the number of newborns delivered in the two tertiary hospitals (7119 newborns in ESUTH, Parklane and 2373 newborns in UNTH, Ituku/Ozalla) could be explained by their location. ESUTH, Parklane is located in the heart of Enugu while UNTH Ituku/Ozalla is located at the outskirts, 15 kilometers from the Enugu metropolis. Pregnant women in labor are more likely to be taken to the tertiary hospital in the city than a tertiary hospital in the outskirts.

In the present study, the prevalence of birth defects was 1.75%, which is comparable to the report of other studies.[11,12] It is interesting to note that a similar study done in Enugu, South East Nigeria, recorded a prevalence of 2.8%.[2] The difference in prevalence rates may be explained by the fact that the current study assessed all the newborns delivered over the 3 years period while the previous study assessed birth defects in already ill babies admitted into the newborn special care unit. One would expect a higher prevalence rate of birth defects in a unit where sick newborns, with possible birth defects, are treated. Cosme *et al.*, in their study of congenital anomaly in Brazil, reported a prevalence of 1.6%.[13] Birth defects affect 3% of live births in the United States of America and 2.55% in Europe.[14,15] These high prevalence rates may be explained by the detailed and thorough search for birth defect through extensive investigations, which was not performed in the present study.

In the current study, males were more affected with birth defects than females. Other studies also recorded the predominance of males affected by birth defects when compared to females.[13,16,17] However, other studies concluded that birth defects are more common in females.[1,5,9] Some authors have postulated that females are more afflicted with lethal birth defects.[6] The exact reason for the gender difference is unknown, but one report suggested the role of p53 inactivation of the X chromosome.[18] Ambiguous genitalia was reported in 0.6% of our patients that had birth defects. This is consistent with the finding of Mashuda *et al.*[9]

Birth defects are mostly detected within the first 1 month of postnatal life.[19] This is in line with the finding in the present study. The severity of birth defects may determine when it is detected. There were more birth defects in preterms. Honein *et al.*, in their study, reported that birth defects are over five times more likely among preterm births when compared with term birth.[20] This may explain why there are more birth defects detected in preterms. In the present study, most of the newborns that had birth defects weighed <2.5 kg at birth. This is in agreement with a study done in Ethiopia.[21] This may be due to intrauterine growth retardation that may be associated with birth defects.[22] However, a report from Tanzania concluded that birth defects are more common in newborns whose birth weights are 2.5 kg and above.[9] In the present study, most of the mothers whose babies had birth defects were aged 20–34 years. Dutta and Chaturvedi documented a statistically insignificant association between maternal age and birth defects, whereas Suguna Bai *et al.* reported a higher incidence of birth defects in babies born to mothers aged over 35 years.[23,24]

There is no conclusive evidence regarding the most appropriate mode of delivery for foetus with birth defects when the prenatal diagnosis is made. Trauma to the foetus can occur following any route of delivery.[25] About 85% of our patients were delivered vaginally. For mothers with the prenatal diagnosis of birth defects, the caesarean section should be performed based on appropriate maternal indications, appropriate foetal indications or maternal request.

With regard to the pattern of birth defects in the present study, the most common system involved was the musculoskeletal system (45.2%). Other studies conducted by other researchers also found the musculoskeletal system as the most common system affected by birth defects.[6,23,26] However, some other studies reported central nervous system abnormality as the most common birth defect.[1,27] Suguna Bai *et al.*, in their study, concluded that birth defects affecting the gastrointestinal tract as the most common birth defect.[24] These differences are difficult to explain. However, the fact these studies were carried out in different geographical areas and at different times may explain it.

Prenatal diagnosis of birth defects is a routine practice in developed countries.[28] However, only about one-tenth of our patients had a prenatal diagnosis of their birth defect. Poverty, ignorance and non-availability of expertise and experience required for interpretation of such ultrasound scan may account for a low level of birth defect detection rate. The lack of experienced radiologists on maternal ultrasound may also explain the high rate of missed birth defects during the maternal ultrasound scan. The benefits of prenatal diagnosis lie in the emotional/financial preparation of the parents and the choice of hospital for delivery, where immediate treatment can be offered to the newborn on delivery. Although the exact pathogenesis of birth defects is unclear, about 2–10th of the mothers in the index study, were diabetics and hypertensives. Pre-gestational diabetes is one of the leading known causes of birth defects

with up to 9-fold increase in birth defects when compared with the rate seen in non-diabetic mothers.[29] Bellizzi *et al.*, in their study, reported that maternal hypertension exposes newborns to a significant risk of developing birth defects, and this risk is exacerbated by superimposing eclampsia.[30]

Initial/immediate care of neonates with birth defects

Immediate care offered to the neonates depended on the type of birth defect. For instance, in intestinal atresia, the neonate was placed in a radiant warmer to avoid hypothermia, nasogastric tube and urethral catheter were passed. The neonate was also placed on glucose-containing intravenous fluid, intravenous antibiotics and nil per oral maintained. In cases of myelomeningocele or congenital talipes equinovarus, other surgical subspecialities were invited to take over management.

Parental counselling

Having a baby with birth defect can negatively impact on the physical and mental health of the parents. There is a need for parental counselling in terms of support groups, surgical repair and prevention such as folic acid supplementation in neural tube defects.

Limitations of this study

Although this was a prospective study, it was limited by the small number of birth defects. A larger number of cases would have availed better analysis.

Only obvious and observable birth defects were assessed for this study. Investigations were not done. Stillborns were excluded from this study.

This study was hospital based. A community-based study would have captured more birth defects.

CONCLUSION

The findings of this study indicate that birth defects are relatively common and diverse. This study has highlighted the prevalence of birth defect of 17.5 cases per 1000 live births in Enugu, Nigeria. The musculoskeletal system is the most affected system. There is a need for a registry system of birth defects and a large community-based study should be conducted in Enugu to determine the incidence of birth defects and their associated factors.

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Conflicts of interest
There are no conflicts of interest.

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