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Congenital anomalies: Prospective study of pattern and associated risk factors in infants presenting to a tertiary hospital in Anambra State, South-east Nigeria

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Abstract: *Background:* Contemporary understanding of the pattern of congenital anomalies is both important in its clinical management as well as in improving the overall health of the community.

Methodology: All infants presenting from January to December 2102 at the centre were prospectively studied.

Results: A total of 5010 infants were screened, of which 108 have congenital anomalies giving a prevalence of 2.2%. Major anomalies were noted in 101 (93.5%) infants. Only two (1.9%) were preterm. Consanguineous relationship was observed in 2 (1.9%) cases. Pre natal Ultrasound scan was done in 33(30.8%), but in only 7 (6.5%) was any anomaly detected

Onitsha, a densely populated urban area contributed 45(42%) of the cases. Only 7(6.5%) of the

mothers took herbal prescriptions in the first trimester of pregnancy. Risk of anomaly progressively rises, reaching a peak of 48% in the maternal age group of 26-30 years and drops steadily after. Gastrointestinal tract anomalies were most common (no. =28, 25.9%). Residences close to dump sites, telecom masts/base stations, electricity cables, industries and heavy motorised highways contributed to risk factors in 9-12 % of cases.

Conclusion: More of younger mothers were affected with gastro intestinal malformations predominating. The location of home environment is a potential risk factor that will require further characterisation.

Key words: Congenital disorder, Malformations, Birth defects, Risk factors, Epidemiology, Nigeria, Anambra

Introduction

Congenital anomalies also known as birth defects, 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 Understanding of the pattern of congenital anomalies is both important in the clinical management of the condition as well as in improving the overall health of the community.

The epidemiology of congenital anomalies varies from period to period and from region to region. In developed economies, the prevalence varies from 1.07% to 4.4%.^{2,3} Establishment of congenital anomaly registry or surveillance has enabled better monitoring of epidemiological trends and institution of preventive mechanisms in some of these nations.^{4,5} In Nigeria, available reports have prevalence rates ranging from 0.75%-13.9%.⁶⁻¹⁰

The aetiology of congenital anomalies is unknown in about 50% cases. Identifiable causes include single gene defects(6-7%), chromosomal abnormalities(6-7%), multi-factorial disorders which are the result of interaction between genetic predisposition and presumed environmental factors(20-25%), and teratogenic factors (6-7%).¹¹ Responsible for the high rate of anomalies in low and medium income countries(LMIC's) is a combination of factors including adverse environmental attributes like high infective diseases, urbanization with attendant slums, pollution, industrial wastes , living close to dump sites , and poor nutrition.¹

The LMIC's also bear the greater burden of increasing perinatal deaths from congenital anomalies estimated at 12.7%.¹² Survivors many times may have lifelong disabilities with impairment of quality of life.

This article aims to study the contemporary epidemiology and risk factors associated with congenital anomalies

lies in our region, Anambra state, south east Nigeria, thus enabling data for establishment of better control measures .

Methodology

Every infant that presented for treatment at the outpatient, inpatient, special baby care and the children emergency departments of the hospital within the study period was screened for the presence of any structural congenital anomaly.

Once a case was identified, a trained investigator further interviewed the parents and examined the infant, obtaining required data using a structured close ended interviewer administered questionnaire. Data was collected for one year (January to December, 2012) and covers bio-data, malformation characteristics, parental attributes, and environmental conditions at the place of habitation.

Ethical approval was obtained from the institutions Ethical Committee. Consent from the child's parent or guardian was also obtained.

Congenital cardiac anomalies were excluded from the study due to unavailability of diagnostic tools.

Study place

Nnamdi Azikiwe University Teaching Hospital is a tertiary hospital in Anambra State, South East Nigeria. Being the only institution in the state with neonatal, paediatric and paediatric surgical services, it receives such patients from all parts of the state and some neighbouring states. It thus serves a population of over five million people.¹³ Anambra is one of the most urbanized states in Nigeria; over 60% of its population live in urban areas with most engaging in commercial activities as occupation.¹⁴

Distances

For the purposes of the study, living within the under mentioned distances is described as living close to the site: 100metres circumference from a refuse dump site, 300metres circumference of a telecommunication mast/base station or a heavily motorised highway and 50metre from high voltage electricity transmission cables. Distances were estimates obtained by respondents guided by trained interviewer.

Data analysis

Data were analyzed using Statistical Package for Social Sciences (IBM SPSS statistics for Windows version 21.0 Armonk, NY: IBM Corp). 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 of < 0.05 .

Results

Of the total of 5010 infants screened, 108 have congenital anomalies giving a prevalence of 2.2%. More males were affected, 71 (65.7%) than females, 34(31.6%) with an M: F ratio of 2:1. Three (2.8%) neonates have Disorders of Sex Differentiation (DSD). Major anomalies were noted in 101(93.5%) infants while the rest 7(6.5%) were minor. Fourteen (13.0%) had multiple anomalies. Only two (1.9%) were preterm. Consanguineous relationship was observed in another 2(1.9%) cases. Prenatal Ultrasound was done in 33(30.8%), but in only 7 (6.5%) was any anomaly detected

Risk of anomaly progressively rises, reaching a peak of 48% in the maternal age group of 26-30 years and drops thereafter. Mean peak maternal age was 8.5years (SD =6.05). Figure 1 shows the trend and compares between maternal and paternal age group occurrences.

Table 1 shows a summary of identifiable environmental risk factors. Twelve infants (11.1%) have associated identifiable multiple risk factors. Only six(5.6%)mothers had any form of chronic illness while 23(24.1%) reported abuse of drugs as shown in tables 2 and 3 respectively.

Table 1: Frequency of identified risk factors

No. of risk factors	frequency	%
None	65	60.2
One risk factor	31	28.7
Two risk factors	7	6.5
Three risk factors	4	9
> 3 risk factors	1	0.9
Total	108	100.0

Fig 1: Trend and comparison between maternal and paternal age groups, with frequencies

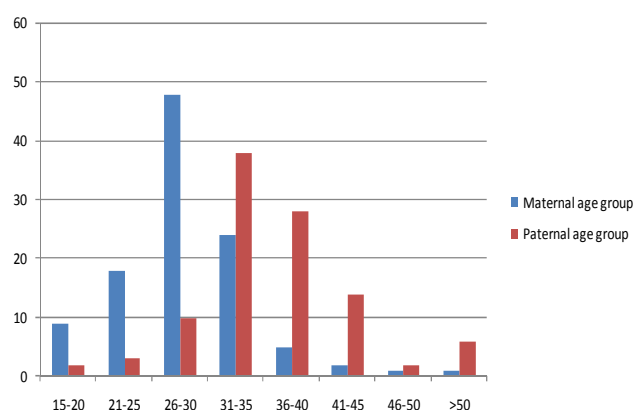
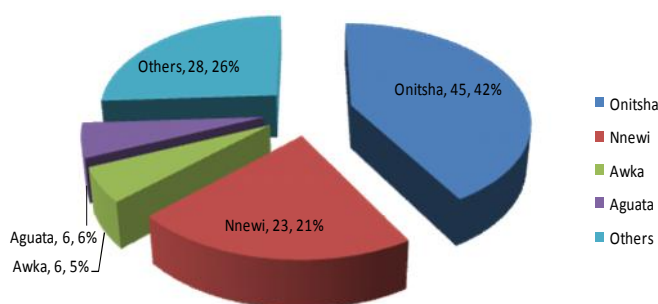


Table 2: Maternal chronic illness during pregnancy

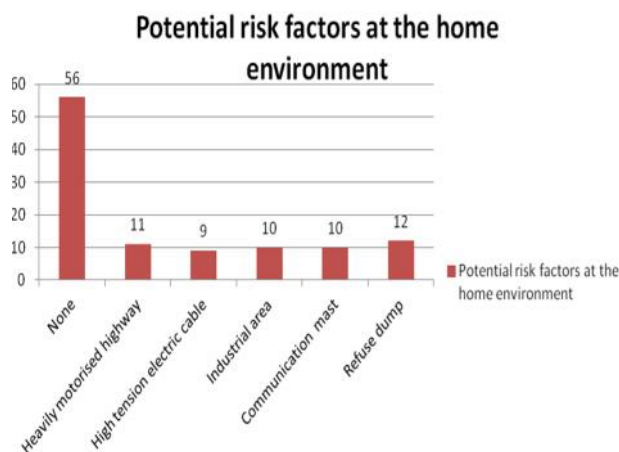
Illness	frequency	%
Diabetes Mellitus	1	0.9
Human Immune deficiency virus	1	0.9
Multinodular goitre	1	0.9
High Blood Pressure	3	2.8
Total	6	5.6

Fig 2: Number of occurrences according to residence**Frequencies of place of residence****Table 3:** Frequencies of drug abuse

Drug ingestion at 1st Trimester	No (%)
Self medication with orthodox drugs	13 (12.1)
Took herbal preparations	7 (6.5)
Alcohol ingestion	6 (5.6)
Cigarette/marijuana smoking	0 (0)

Table 4: Distribution of the anomalies

Type of anomaly	Frequency	%
<i>Central Nervous System</i>		
Spina bifida	8	7.4
Hydrocephalous	6	5.5
Encephalocoele	2	1.9
Scalp dermoid	1	0.9
Microcephalous	1	0.9
Total	(18)	(16.7)
<i>Gastrointestinal tract</i>		
Intestinal atresias	8	7.4
Omphalocoele	6	5.5
Anorectal malformation	6	5.5
Hirschsprung's disease	3	2.8
Gastroschisis	2	1.9
Malrotation	2	1.9
Oesophageal atresia	1	0.9
Total	(28)	(25.9)
<i>Urogenital System</i>		
Hypospadias	10	9.3
Posterior Urethral Valve	7	6.5
Disorders of sex development	3	2.8
Prune belly syndrome	2	1.9
Bladder extrophy	1	0.9
Epispadias	1	0.9
Total	(25)	(23.1)
<i>Musculoskeletal</i>		
Polydactyly	5	4.6
Congenital Talipes EquinoVarus	5	4.6
Syndactyly	2	1.9
Aplasia cutis congenita	1	0.9
Total	(13)	(12.0)
<i>Face</i>		
Cleft lip/palate	15	13.9
Microtia	1	0.9
Haemangioma	3	2.8
Total	(19)	(17.6)
<i>Tumours</i>		
Cervical teratoma	2	1.9
Sacrococcygeal teratoma	1	0.9
Total	(3)	(2.8)
<i>Chromosomal</i>		
Down's syndrome	3	2.8
Total	(3)	(2.8)
Grand total	108	100/

Fig 3: Potential risk factors at the place of residence**Discussion**

The prevalence of 2.2% from this study closely resembles rates from other reports from the South Eastern region of the country.¹⁵ It is however lower than rates from the northern part of the country (13.9%) where adequate antenatal care utilization is reportedly low.¹⁰ About 90% of the mothers in our study received antenatal care from the second trimester. It may be adduced that an improved education, improved socioeconomic state and an increased awareness may have influenced this higher health seeking behaviour and hence a lower prevalence.

However, despite this high antenatal care rate, our study showed both low level utilization of prenatal diagnosis and low capacity of identifying positive cases. Only about a third (33%) of the study population with congenital malformation had prenatal ultrasound. Abnormality was only detected in 6% of these. Prenatal ultrasound has an overall anomaly detection rate of 35%.¹⁶ The low sensitivity may be attributed to the fact that more than 90% of these screening were not done in a tertiary centre. Sensitivity, specificity and positive predictive value are better when high risk pregnancies are screened in a tertiary hospital.¹⁶

The pattern of anomalies varies from period to period and from region to region. In our study, anomalies of the gastrointestinal tract had the highest occurrence of 28 (25.9%), followed in order by the urogenital tract (23.1%) and the central nervous system (16.7%) (Table 4). The predominance of gastrointestinal anomalies was also documented by several other Nigerian reports.^{10, 17} However few other centres have central nervous system or musculoskeletal system as the most common affected system. Of the individual anomalies; cleft lip/palate (13.9%), hypospadias (9.3%), atresias (7.4%) and Spina bifida (7.4%) were the leading anomalies. The high rate of cleft lip/palate anomalies may be attributed to the hospital being recently adopted as a partner for the *Smile Train* (a United State based international children's charity) free corrective repairs programme and thus was receiving referrals from many other regions.

While the maternal age group with the highest number of occurrence was 26-30 years, 75% of the mothers were aged 30 years and below (Figure 1). Prevalence was higher in young mothers and progressively rises, peaking at 30 years and then declines steadily. Twenty eight percent of the mothers were primipara. However there was no significant relationship between age of primip with the type of the anomaly ($p = 0.633$). The high number of young mothers with congenital anomalies can be explained from the high number of structural, non chromosomal anomalies in our study in contrast to chromosomal anomalies which increases with increasing maternal age.¹⁸

We recorded only 1.9% preterms. This low level may be alluded to the fact that majority of the babies were delivered outside the tertiary centre. It may be possible that some affected neonates may have not survived to reach the tertiary hospital. Also the study did not include still births and early prenatal deaths. Only two cases of consanguineous relationship were identified. This is understandable as in our setting consanguinity is not practised and is viewed as a taboo. The two reported cases were actually infants from northern Nigerian nomadic Fulani settlers.

Of the major cities where the patients reside in, Onitsha contributed almost half (42%) of the burden of congenital anomalies prevalence, followed by Nnewi at 21% (Figure 2). These cities, especially Onitsha, are densely populated urban cities in Anambra state with high vehicular traffic, industrial pollutions and their share of urban slums. The high prevalence may be related to the dense population, it is also possible that certain environmental factors as stated earlier may be responsible for the increase. Most common anomalies from these areas were urogenital (no=13, 12.0%) and gastrointestinal (no =9, 8.3%). Further studies will be needed to fully understand the specific role of environment on the high rate of anomalies on children born in these areas.

Worldwide more than half of the aetiology of congenital anomalies is unknown. Environmental factors either alone or in conjunction with genes account for about

40% of the aetiology. In our study about 9-14 % of the mothers while pregnant were found to have been exposed to either one or a combination of pollution from heavy vehicular traffic, industries, or living near a refuse dump, high tension electricity transmission cable or telecommunication mast / base stations (Figure 3). Concerns have grown on the effect of these modern conveniences on the reproductive health. Studies are quite scanty in this area. Living near telecommunication base stations or masts, which can transmit 1900-2200MHZ frequency range, can expose one to high levels of radiofrequency waves which can affect developing foetus.¹⁹ Living close to a highway predisposes one to pollutants like carbon monoxide, hydrocarbons and particulate matter all of which have adverse reproductive outcomes.²⁰ High electromagnetic fields from power lines, pollutants from industries and emissions from dump sites all have the potential of negatively influencing birth outcome. More targeted studies however are still needed to understand better the roles of these risk factors.

Conclusion

In conclusion, the prevalence of congenital anomalies in Anambra state is low, but with concentrations in urban areas with high population densities. Living close to dump sites, telecom masts/base stations, high voltage electricity cables and heavy motorised highways are potential risk factors that will need further specific studies for characterisation.

Limitations

Even though the data was collected from the only hospital serving an entire state; being a hospital based study, the results may not have been an exact reflection of the pattern in the general population. A community based study will better assess the true pattern.

Conflict of interest: None

Funding: None

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