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Research Article

Profile of Prenatally Diagnosed Major Congenital Malformations in a Teaching Hospital in Nigeria

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Abstract

Background: Prenatal diagnosis of major congenital abnormality is one of the main goals of antenatal care, because of its contribution to perinatal morbidity and mortality. Awareness of the profile in terms of rates and spectrum could aid management and prevention strategies. This study aims to determine the profile of congenital malformations, and the relationship between the **rates** and some maternal socio-demographic and obstetric variables.

Methods: A retrospective cross-sectional study of prenatally diagnosed congenital malformations in singleton pregnancies over a four-year period. The ultrasound scan findings and the findings of fetal ultrasonography, together with maternal socio-demographic and obstetric variables, were collected from the ultrasound scan reports or medical records of each pregnancy. Data were analyzed using Microsoft Excel 2010.

Results: Among the 968 singleton pregnancies, 78 had major congenital malformation, giving an antenatal rate of 8.04/1000 (0.8%). The first trimester prevalence was comparable with other trimesters. Malformation mostly involved single systems (93.6%), which are mainly central nervous (48.7%) and gastrointestinal/abdominal systems (21.8%). The rate was statistically significant (<0.0018) in women aged >35 years. The mean maternal age and parity were 31.4 + 4.7 and 2.8 + 0.4, respectively. The rates of congenital malformation in spontaneously or assisted conceptions were not statistically significant (p = 0.073 and p = 0.085).

Conclusion: Maternal age >35 years and multiparity are important risk factors for congenital malformation. The commonly involved systems are the central nervous and gastrointestinal systems.

Key words: Congenital malformations; prevalence; spectrum; antenatal ultrasound scan; Nigeria

Introduction

Congenital malformations (CM) are structural or functional anomalies that occur during intrauterine life and can be diagnosed in the prenatal period, at birth or later in life [1].

It is an important cause of perinatal morbidity and mortality, with an estimated 303,000 neonatal deaths occurring within four weeks of birth every year, worldwide [1–3]. CM are commonly diagnosed in late pregnancy, during the neonatal period, or during infancy [2, 4–6]. However, the rise in the rate of diagnosis in the first trimester can be attributed to the widespread availability and utilization of prenatal ultrasound scan [5].

Studies on the prevalence and the spectrum of major CM are mainly from developed countries, leaving only few published studies from developing countries, especially in Nigeria. Good epidemiological data on the rate and pattern of CM in a specific region provides the opportunity to identify possible etiological factors and can be useful for their prevention in this country and across the wider region [7]. The preventive measures include vaccination, folic acid and iodine supplementation, and reduction in alcohol intake can also be adopted against common malformation in an environment [1].

The primary aim of the study was to evaluate the spectrum of major CM, while the secondary aim was to determine the relationship between CM and some maternal sociodemographic and obstetric variables.

Methods

This is a retrospective cross-sectional study of the ultrasound scan or medical records of pregnant women, who had ultrasound scan over a four-year period (July 1st, 2014–June 31st, 2018), in the fetal medicine or radiology unit of the Olabisi Onabanjo University, Sagamu, Ogun State Nigeria. Ultrasound scans were performed by a Fetal-Maternal Medicine specialist certified by Fetal Medicine London to perform anomaly scan and a Consultant Radiologist with experience in ultrasound diagnosis of fetal anomalies. The scan machines used were the Voluson P8, BT 2005 (GE Kretz, Austria) or Phillips HDI 1500. Maternal socio-demographic and obstetrics parameters were extracted from ultrasound scan reports and medical records. Only major CM in singleton pregnancies were included for analysis, while minor CM were excluded. Major structural anomalies are defined as structural changes that have significant medical, social, or cosmetic consequences for the affected individual, and typically require medical intervention. Major CM is defined according to the standard anatomical nomenclature of the system(s) involved. They are classified as single or isolated if only one body system is involved

and complex or multiple, if >2 systems are involved. The overall antenatal rate of CM was calculated from the ratio of number of fetuses to the CM and the total number of pregnancies scanned.

Data was analyzed using 2010 Microsoft excel. Descriptive data are expressed in percentages and presented in simple frequency tables. Mean and standard deviations were used as where appropriate. The statistical significance of the data relationship was defined using $p < 0.05$.

Results

A total of 9,698 women had prenatal ultrasound scans during the study period, of which 853 (8.8%) were in the first trimester, 5,394 (55.6%) in the second trimester, and 3,453 (35.6%) in the third trimester; 78 out of the 9,698 fetuses (0.8%) had congenital abnormalities. The malformed fetuses constitute the primary subjects of this study (Table 1). Most of the malformations were diagnosed in the second (55.6%) and third (35.6%) trimesters, while only 8.8% were diagnosed in the first trimester.

TABLE 1: Frequencies of Major Congenital Malformations During Pregnancy.

Gestational Period (Trimester)	Number of Pregnancies	Frequencies (%)	Number of Congenital Malformations	Frequencies (%)
First Trimester	853	8.8	6	0.7
Second Trimester	5,394	55.6	41	0.8
Third Trimester	3,451	35.6	26	0.8
Total	9,698	100.0	78	0.8

The spectrum of diagnosed congenital abnormalities is shown in Table 2. Central nervous system malformations were diagnosed in 38 (48.7%) fetuses, with the commonest being hydrocephalus or ventriculomegaly in 22 (57.9%) fetuses and acrania or anencephaly in 13 (34.2%) fetuses. Holoprosencephaly was diagnosed in 2 (5.3%) fetuses and Dandy-Walker Malformation in only 1 (2.6%) fetus. Gastrointestinal system malformations were found in 17 (21.8%) fetuses, with 7 (41.2%) presenting with polyhydramnious. Polycystic kidney was the commonest (57.1%) urogenital system malformation, followed by lower urethra tract obstruction (LUTO) in 28.6% and hydropelvis in 14.3%, respectively. Malformations of the musculoskeletal system

Table 2: Spectrum of Major Congenital Malformations.

Systems	Congenital Malformations	Number (%)
Central Nervous System 38 (48.7%)	Hydrocephalus/Ventriculomegaly	22 (57.9)
	Acrania/Anencephaly	13 (34.2)
	Holoprosencephaly	2 (5.3)
	Dandy Walker Malformation	1 (2.6)
Gastrointestinal System 17 (21.8%)	Duodenal Atresia	7 (70.0)
	Jejunal Atresia	1 (30.0)
	Gastroschisis	3 (30.0)
	Omphalocele	6 (60.0)
Urogenital System 14 (17.9%)	Lower Urethral Tract Obstruction	3 (21.4)
	Hydropelvis	2 (21.4)
	Polycystic Kidney	8 (57.1)
Musculoskeletal System 13 (16.7%)	Thanatophoric Dwarfism	5 (38.5)
	Skeletal Dysplasia	8 (61.5)
Cardiovascular System 2 (2.6%)	Tetralogy of Fallot	1 (50.0)
	Ventricular Septal Defect	1 (50.0)
Respiratory System 1 (1.3%)	Congenital Cystic Adenomatoid Malformation	1 (100.0)
Systems Involved		
Multiple (>2 systems) 73 (93.6%)	–	5 (6.4%)
Single (1 system) 5 (6.4%)	–	73 (93.6)

were found in 13 fetuses (16.7%) cases. Two fetuses (2.6%) displayed cardiovascular malformations: tetralogy of Fallot (TOF) and ventricular septal defect (VSD) were diagnosed. The VSD was postnatally confirmed, while the TOF ended in miscarriage. Respiratory system abnormality was the least prevalent malformation diagnosed in the study (1.3%). The malformation involved one system in 73 (93.6%) fetuses and multiple systems in 5 (6.4%) fetuses

The maternal age in the study ranged between 21 and 52 years, with a mean of 31.5 ± 4.7 years. The mothers above the age of 35 years were 3,446 (35.5%); 47 (53.8%) out of the 78 fetuses with CM were in this category ($p \leq 0.0018$). The mean and range of parity were 2.8 + 0.4 and 0–8, respectively; 52 (66.7%) fetuses with CM were diagnosed in mothers who were para ≥3. Pregnancy was spontaneously conceived in 8,139 (83.9%) women, among whom were 74 (94.5%) fetuses out of all malformed fetuses ($p = 0.073$). Conception was achieved by assisted reproductive technology in 1,559 (16.1%) women, and 5.1% ($p = 0.085$) of all the fetuses with CM were in this group (Table 3).

TABLE 3: Relationship of Major Congenital Malformations with Maternal Sociodemographic and Obstetric Variables.

Maternal Sociodemographic and Obstetric Variables	Range	Number		Frequencies (%)	*p* value
		No (n = 9,698)	No CM (n = 78)		
Maternal Age	20–25	1,130	4	0.4	0.076
	26–30	2,007	5	0.7	
	31–35	3,115	22	0.7	
	36–40	1,284	39	3.1	0.0018
	41–45	2,028	6	0.3	
	>46	134	2	1.5	
Parity	0–2	5,237	26	0.4	0.0056
	3–4	3,992	44	1.1	
	>5	429	8	1.7	
Mode of Conception	Spontaneous/ Natural	813,974		0.9	0.073
	Assisted	15,594		0.3	0.085

Discussion

Prenatal diagnosis of CM is one of the key goals of antenatal care. In the study, 0.8% of fetuses had CM. The prevalence of major CMs range between 0.6% in Barbados [7] and 2.8% in Saudi Arabia [4], and 23.9 per 1,000 births for 2003–2007 in Europe [8]. In Nigeria, the prevalence reported is between 2.7 and 6.2% [9, 10]. The prevalence from our study is strikingly lower than rates reported from other local studies. The variations could be due to differences in study population characteristics, presence of environmental teratogens, local sociocultural factors that encourage concealment of information about birth defect and study design among others. The highest rate of 6.2% was reported in a teaching hospital study where the environment was affected by oil spillage and its consequent environmental degradation effects. Crude oil spillage is believed to have teratogenicity effects on the fetus, which could explain the higher rates of occurrence of CM in the pregnant population. In contrast, a lower rate was reported in other areas that are not affected by oil-induced environmental degradation. Consanguinity and culture of concealment of disease data may explain the difference between our study finding from some other environments in the Middle East and Europe. In Omani population, about 2.5% of the babies at birth were affected by CM compared with the 1.5% in non-Omani births ($p < 0.05$) [11].

The profile of first-trimester prenatally diagnosed CM is relatively scarce in scientific literature. The practice should however be encouraged because it provides opportunity for early counseling and fetal therapy [4]. Late pregnancy booking and scarcity of experts in first trimester diagnostic scans could contribute the present situation. It is however instructive to observe that while the lowest proportion of all antenatal ultrasound scans was in the first trimester, the proportion of CM cases diagnosed in this first trimester to number of ultrasound scans was similar to other trimesters. These findings show that the first trimester is an equally important period to diagnose fetal anomalies, which could have resulted in abortion before the time of second trimester scan. The predominance of CNS abnormality over abnormalities of other systems was also confirmed in our study [9, 10, 12]. The relatively larger size of the fetal head is believed to explain the relative ease of diagnosis due to easy visualization of its structures. The cardiovascular system anomaly was in the lowest data range, similar to the reports from other studies [4, 13].

The relationship between CM and maternal age was shown to be unimodal, contrary to the bimodal distribution [14]. The risk for CM in women aged >35 years can be explained by factors such as the high risk of maternal age-induced aneuploidy, and increased incidence of medical complications such as diabetes mellitus could be responsible for the unimodal distribution [14]. Multiple systems abnormality is commoner in regions with higher risk of consanguineous marriage [14].

Conclusion

This study shows that profiling CM is similar to many other hospital-based studies. The first trimester ultrasound data should encourage a policy for routine first trimester scanning in women that present in early pregnancy, while routine second trimester scanning should be routinely done in all other pregnancies. Furthermore, postnatal confirmation of all antenatal diagnosis should be done as an important component of auditing. A multicenter study that would give a better representative population rate is recommended to be done by expert sonographers.

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Conflict of Interest

The authors declare that there is no conflict of interest.

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