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A Study on the Incidence of Congenital Musculoskeletal Anomalies in a Tertiary Hospital in Enugu, Nigeria

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Review Article

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Abstract

Background: Congenital musculoskeletal anomalies refer to anomalies affecting the skeletal and muscular systems present when the child was born. In Nigeria, congenital anomaly of the musculoskeletal system is rarely reported. The aim of this study was to determine the incidence and distribution of clinically obvious congenital musculoskeletal anomalies among live born neonates at a teaching hospital in Enugu, Nigeria.

Materials and Methods: This was a retrospective study carried out on neonates at a teaching hospital in Enugu. The newborns were examined for congenital musculoskeletal anomalies and the study covered a period from 2016 to 2020.

Results: There were 7, 011 babies delivered in the teaching hospital during the study period. Overall, 109 babies had at least one congenital anomaly which gave an overall incidence of all congenital anomalies of 1.6% or 16 babies per 1000 live births. Out of the 109 babies that had congenital anomalies, 49 babies had a musculoskeletal anomaly. This gave an incidence of 0.7% or 7 babies per 1000 live births. There was male predominance and about half of the babies were delivered preterm. Syndactyly and polydactyly were the most common congenital musculoskeletal anomalies. Majority of the mothers were older than 35 years and were delivered vaginally. Two-thirds of the mothers had maternal illness during pregnancy and one-tenth had a previous history of congenital anomaly.

Conclusion: Congenital musculoskeletal anomalies are seldom reported. Polydactyly, syndactyly and talipes are the commonly seen anomalies.

Keywords: Congenital, birth defects, musculoskeletal, tertiary hospital.

Introduction

Congenital anomalies or malformations, also known as birth defects, may be defined as structural defects present at birth which may lead to physical and mental disabilities [1]. Congenital anomalies represent defects in morphogenesis during early fetal life [2]. In developing and developed countries, congenital anomaly is a significant cause of infant morbidity and mortality. Although in poorer countries, malnutrition and infections take prominence [2]. In Asia, congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths [1,3]. Congenital musculoskeletal anomalies refer to anomalies affecting the skeletal and muscular systems present when the child was born. Multiple congenital anomalies can occur in a neonate and present a difficult challenge to the clinician [2]. The etiology of congenital anomalies is believed to be multifactorial attributable to faulty development due to complex interaction of genetic and environmental factors [1]. Exposure to various teratogens during intrauterine life may affect the developing fetus resulting in permanent postnatal damage. Some of the teratogens include radiations, thalidomide and certain infections such as rubella [1]. Maternal age, parity, and antenatal illness are associated with congenital malformations [4]. The overall incidence of the entire spectrum of congenital anomalies has been quoted as 2% of total births [5]. Some studies have reported congenital anomalies of the musculoskeletal system as the most commonly [6,7]. Available data on congenital musculoskeletal anomalies in sub-Saharan Africa are sparse. This study was an attempt to determine the incidence and distribution of clinically obvious congenital musculoskeletal anomalies among live born neonates at a teaching hospital in Enugu, Nigeria.

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Materials and Methods

This was an observational study carried out at Enugu State University Teaching Hospital (ESUTH), Enugu, Nigeria. The hospital serves the whole of Enugu State, which according to the 2016 estimates of the National Population Commission and Nigerian National Bureau of Statistics, has a population of about 4 million people and a population density of 616.0/km2. The hospital also receives referrals from its neighboring states. Ethical approval was obtained from the ethics and research committees of the hospitals and informed consent was obtained from the patients' mothers. For the purposes of this study, our interest was on clinically obvious and observable abnormality of musculoskeletal system, which was noticed at birth or a few days after birth. All the live babies born in ESUTH during the period of this study were included. Stillborn were excluded from this study. This study covered a period of 5 years, from January 2016 to December 2020.

The protocol of this study conforms to the declaration of Helsinki. All the consecutive children who were born in ESUTH during the study period had a thorough physical examination (general and systemic) performed by a pediatrician and a pediatric surgeon at birth and at 6 weeks postnatal visit. This 2-stage physical examination minimized the error of missing any congenital anomalies of the musculoskeletal system. Diagnosis of congenital anomalies of the musculoskeletal system was based only on clinical evaluation of the babies by the pediatrician and pediatric surgeon. Investigations such as radiography, ultrasonography, and computed tomography scan and magnetic resonance imaging of the skeletal system were not performed.

For each patient, the following data were collected: 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 kilograms (kg) were considered to be normal while birth weight less than 2.5 kg were considered as low birth weight. Babies born at less than 37 completed weeks, calculated from the first day of last menstrual period, were considered preterm while babies born at or after 37 completed weeks were considered term. Other data collected include family history of congenital anomaly, maternal comorbidities such as diabetes/hypertension and use of herbs in early pregnancy.

Data Analysis:

Statistical Package for Social Science (SPSS) for Windows version 23 (IBM Corp., Armonk, NY) was used for data entry and analysis. Data were expressed as percentages, medians and means

Results:

Patents' demographics:

A total of 7, 011 babies were delivered during the study period, of whom 109 had at least one congenital anomaly. This gives an overall incidence of all congenital anomalies of 1.6% or 16 babies per 1000 live births. Out of the 109 babies that had congenital anomalies, 49 babies had a musculoskeletal anomaly. This gave an incidence of 0.7% or 7 babies per 1000 live births. Congenital anomalies of the musculoskeletal system accounted for 38.9% of all the systems.

There were 31 (63.3%) males and 18 (36.7%) females. Twenty-eight (57.1%) babies were term and 21 (42.9%) were preterm babies. Twenty-five (51%) babies weighed 2.5 kilograms and above while 24 (49%) babies weighed less than 2.5 kilograms.

Distribution of congenital musculoskeletal anomalies (n=49):

This illustrated in Table 1.

Table 1: Pattern of the congenital musculoskeletal anomalies.

Congenital anomalies	Number of patients	Percentage
Syndactyly/Polydactyly	14	28.5
Talipes (club foot)	6	12.2
Pes planus (pes planus)	5	10.2
Pectus carinatum/excavatum	4	8.2
Rocker Bottom foot	4	8.2
Congenital Scoliosis/Kyphosis	4	8.2
Webbed toes	4	8.2
Amelia/Phocomelia	2	4.1
Arachnodactyly	2	4.1
Craniofacial abnormalities	2	4.1
Prune belly syndrome	1	2
Aphalangia	1	2

Maternal Parameters:

The possible maternal variables are shown in Table 2.

Table 2: Profile of maternal parameters.

Maternal factors	Number of Patients	Percentage
Maternal age		
Less than 35 years	11	22.4
35 years and above	38	77.6
Mode of delivery		
Vaginal	31	63.30%
Caesarian section	18	36.70%
History of maternal illnesses e.g. diabetes, hypertension		
No	33	67.3
Yes	16	32.7
History of previous congenital anomaly/consanguinity		
No	44	89.8
Yes	5	10.2
Use of herbal concoction		
No	28	57.1
Yes	21	42.9

Prenatal diagnosis:

Four (8.2%) patients had their musculoskeletal anomaly diagnosed prenatally during antenatal ultrasound scan. Two were polydactyly and the other 2 were talipes. Not all the patients had antenatal scan: Some mothers (50%) were unbooked. In addition, some of the congenital musculoskeletal anomalies may have been missed during antenatal scan depending on the experience and expertise of the sonographer. Antenatal anomaly scan is not routinely performed in developing low-income country like Nigeria.

Discussion

Interest in congenital malformations goes back to the dawn of history. William Harvey in his study on the possible causes of congenital malformations documented the teratologic phenomena and disturbance of fetal development [8]. According

to the World Health Organization (WHO) document of 1972, congenital malformation should be confined to structural defect that are at birth [9]. However, in 2012, WHO included structural and functional anomalies including metabolic disorders as congenital abnormalities [10]. Our interest in the present study is on the structural musculoskeletal defects that are present at birth.

The overall incidence of congenital anomalies documented in the current study is comparable to the report of other series on congenital anomalies [10,11]. However, other authors reported lower and higher incidence rates of congenital anomalies [12,13]. The difference in incidence rates may be due to the discrepancies in detailed search and investigations performed for congenital anomalies in the different studies. The specific type of congenital anomaly may also determine its incidence. The incidence of congenital anomalies may vary over time or with geographical location [10].

Congenital musculoskeletal anomalies accounted for about one-third of all the anomalies in the current study and this is consistent with the report of Agrawal et al [1]. One study from Iraq reported musculoskeletal anomaly as accounting for one-fourth of all congenital anomalies [12]. The setting and period of the study may explain these differences. There are temporal and spatial differences in specific types of congenital anomalies. Congenital anomalies of the musculoskeletal system can be as high as 40% of all the anomalies [14].

Syndactyly and polydactyl were the most common congenital musculoskeletal anomalies recorded in the present study. This finding is in agreement with the report of a study from Korea [15]. Howbeit, other studies reported talipes as the most common [4, 10]. Daldrup et al reported dislocation of the hip as the most common congenital musculoskeletal anomaly [16]. The age group of the patients assessed may determine the predominant musculoskeletal anomaly obtained. For instance, the mild form of talipes equinovarus (unilateral postural clubfoot) may not be classically evident at birth. In comparison, polydactyly is easily noticed even by the child's parents.

In the current study, majority of the mothers whose children had musculoskeletal anomalies were older than 35 years. One study from Latvia reported that the prevalence rate of congenital anomalies increased depending on the maternal age [17]. The authors documented that there is higher proportion of preterm births and low birth weights among mothers aged 35 years and above [17]. Ironically, Goetzinger et al reported that advanced maternal age is associated with overall decreased risk for major congenital anomalies [18]. These findings may suggest that the "all or nothing" phenomenon plays a robust role in embryonic development with advancing oocyte age, with anatomically normal fetuses more likely to survive [18]. About two-thirds of the babies with congenital musculoskeletal anomalies, in the index study, were delivered vaginally. Some congenital musculoskeletal anomalies such as polydactyly may not determine the method of delivery. The mode of delivery is determined by obstetric, maternal and fetal concerns.

Maternal diseases such as gestational diabetes mellitus and hypertension may be involved in congenital anomalies. About one-third of the studied patients' mothers had a history of one illness or the other. Giardina et al reported association between the fetus and the mother during intrauterine life; they reported that maternal diseases may have negative impact on the fetus [19]. Maternal diabetes has toxic effects on the development of the embryo and the risk of congenital malformation is increased by three- to four folds [20].

History of previous congenital anomaly/consanguinity is an important consideration when evaluating patients with congenital abnormalities. Ameen et al reported the relevance of maternal history of previous congenital anomalies and the risk of the progeny having congenital anomalies [13]. Most congenital anomalies may be sporadic as only 10% of our patients had a previous history of having had a baby with congenital anomalies.

About half of the mothers whose children had congenital musculoskeletal anomalies took some herbs during their pregnancy. Illamola et al reported the association between the use of herbal medicine during pregnancy and congenital malformations [21]. Laelago reported that most of the women took the herbal medicine in an effort to treat nausea and vomiting [22].

Congenital musculoskeletal anomalies may be diagnosed prenatally. Fordham et al reported that subtle musculoskeletal anomalies become more recognizable in the second and third trimester [23].

Conclusion

Congenital musculoskeletal anomalies are seldom reported. Polydactyly, syndactyly and talipes are the commonly seen anomalies. Gestational diseases such as diabetes should be well treated before and during pregnancy. Intake of herbal concoctions by pregnant women is to be discouraged and antenatal anomaly scan for in-utero detection of congenital anomalies should be encouraged.

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