

## A RETROSPECTIVE STUDY OF CONGENITAL ANOMALIES PRESENTED AT TERTIARY HEALTH FACILITIES IN JOS, NIGERIA

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### ABSTRACT

Early developmental stages are particularly susceptible to disruption because this is the period for organogenesis. This refers to the 4th to 8th weeks of development. The aim of this study was to determine the pattern of congenital anomalies presenting in tertiary health facilities in Jos and any association between the anomalies. A retrospective study of 200 cases of congenital anomalies that presented at the Jos University Teaching Hospital and Plateau State Specialist Hospital Jos-Nigeria was carried out. The data was collected from the Special Care Baby Unit of the health facilities and covered the period between January 1998 and December 2005. Anomalies of the gastrointestinal system had the highest incidence with 61 cases (30.5%); central nervous system abnormalities showed 49 cases (24.5%), skeletal system, 16 cases (8.0%), cardiovascular system, 15 (7.5%), urogenital system 10 (5.0%), chromosomal anomalies, 15 (7.5%), cutaneous system 4 (2.0%), metabolic disorders, 3 (1.5%) whereas 27 cases (13.5%) presented with multiple anomalies. There was no association found between the occurrences of the various congenital anomalies. The Health facilities in Jos metropolis showed various cases of congenital anomalies involving many organ systems. To reduce mortality rate therefore, it is important that careful examination of newborns be carried out in order to detect (and correct) early cases of congenital anomalies.

**Keywords:** *Congenital anomalies, Tertiary Health facilities, Jos University Teaching Hospital, Plateau Specialist Hospital, Special care baby unit.*

### 1. INTRODUCTION

Congenital anomaly is any abnormal structural or medical condition that is present at birth [1]. It is in contrast with the word “acquired”. This can be recognized before birth (prenatal), at birth or even long after birth (postnatal). Congenital anomalies include minor physical defect such as birthmark, severe defects like congenital heart defect and combinations of other abnormalities affecting several parts of the body. Defects of metabolism present at birth are also classified under this medical condition. Congenital anomalies may be inherited or sporadic, isolated or multiple, gross or microscopic [1], [2].

Records of human congenital malformation in cave paintings, sculptures and ultimately in writings date back to prehistoric period. These records have reports of human congenital anomalies such as achondroplasia, conjoined twins, which were often portrayed with mermaids and other fanciful creatures [3].

Congenital anomalies contribute a significant proportion of infant morbidity and mortality as well as fetal mortality. The case fatality rate for most severe anomalies such as anencephaly, trisomy 13 and trisomy 18 and severe heart defects are virtually 100% by the child’s first birthday [4]. They account for 3.3% of all admissions into hospitals and up to 85% of all mortalities in a newborn in Tanzania [5]. In India, it has been observed that they constitute 22% of all early neonatal deaths [6]. In Nigeria the incidence of congenital anomalies associated with central nervous system varies from 1.37/1000 to 5.2/1000 live births [7]. Although it has been reported that out of the approximately 350,000 children born in Canada each year, most are born healthy and at term, about 2-3% of these babies present with serious congenital anomalies [4].

In spite of the frequency of congenital anomalies, the underlying causes for most remain obscure. It has been estimated that around 15%-25% are due to recognized genetic conditions (chromosome and single gene causes), 8%-12% are due to environmental factors (maternal-related conditions, drug or chemical exposures) and 20%-25% are due to multifactorial inheritance [8]. The majority, 40%-60% of congenital anomalies have unexplained causes [9].

The aim of this study is to determine the pattern of congenital anomalies presenting at Jos tertiary health facilities and to establish any inter-relationship. Congenital anomaly remains one of the leading causes of Infant mortality. Thus the place of this research in the programming of a better health care delivery cannot be overemphasized.

This research will provide a database for further studies on causative agents of congenital anomalies in Jos, and help in the suggestion of preventive measures in years to come. When compared with other geographical zones, the findings of this research will be used to compare and contrast findings in other parts of Nigeria.

### **Limitation of study**

Unavailability of data in the majority of the health centers in plateau state is a major limitation to this research. Of all the health institution in plateau state, only Jos University Teaching Hospital (JUTH) and Plateau State Specialist hospital Jos, have a significant record on congenital anomaly as at the time of the study.

Even in health institutions where data are kept, there exists the problem of poor management of data due to incessant strike and other internal problems. This causes low and non-uniform data ascertaining cases of congenital anomaly. Many birth defects are not obvious at birth. Those obvious at birth e.g. Cleft palate are more likely to be diagnosed than hidden defects e.g. Renal anomalies and Cardiac defects which may be diagnosed after mother child have left the maternity unit. Prenatal diagnosis of congenital anomalies is yet to be available in Plateau state.

## **1. MATERIALS AND METHODS**

This is a quantitative research method that deals with the systematic collection, analysis and interpretation of data on congenital anomaly in Plateau state. It is an observational research that mainly deals with quantities and frequency. This descriptive study shows the amount (frequency) and pattern of distribution of congenital anomalies among the residents of Plateau state.

### **Variables measured**

- Congenital anomalies of all the biological systems.
- Genetic Disorders
- Metabolic disorders that are congenitally derived

### **Study location**

This study was carried out in Jos, the capital of Plateau state, which is a microcosm of Nigeria, because it contains within itself virtually all the tribes in Nigeria. The state itself has over thirty indigenous ethnic groups. Some of these include Berom, Tarok, Geomai, Ngas e.t.c

Plateau state has a population of about 3,245,750 people by the census of 1991. It is located in the North Central region of the country and lies between latitude 70 and 110 degrees, North and Longitude 70 and 250 degrees East. The landscape is almost treeless. Plateau state shares boundaries with other states like Nassarawa, Bauchi, Kaduna and Taraba [10].

Jos has a mean annual maximum temperature of 80 degrees Fahrenheit, and a mean annual minimum temperature of 62 degrees Fahrenheit. The low relative humidity is less than 25% between November and March. During this period the weather is extremely cold.

### **Study type**

This is a descriptive study, which shows the amount (frequency) and pattern of distribution of congenital anomalies among the residents of Plateau state.

Study design was retrospective in nature by collecting data from Special Care Baby Unit (SCBU) of the Paediatrics Department of Jos University Teaching Hospital (JUTH) and Special Care Baby Unit (SCBU) of Plateau State Specialist Hospital, Jos. All admissions cases (i.e. referral cases and in-hospital birth cases) into the special baby care unit of the two health institutions were taken into consideration.

### **Sample Size and method of collection**

In all, 200 cases of congenital anomalies were recorded during the period. The data were collected from the congenital anomaly registers of the two institutions. Qualified health personnel in the two health institutions properly filled in these registers.

### **Data analysis**

Frequency tables, Pie Charts and Bar charts were used to represent the variables.

### **Statistical test**

The Methodology for the statistical test is **CHI-SQUARE TEST**.

Calculation for the significant difference between the congenital anomalies of the central nervous system and the gastrointestinal system was carried out.

## 2. RESULTS

Out of a total of 200 congenital anomalies recorded, that of the gastrointestinal system was the highest (30.5%), followed by the central nervous system with an occurrence of 24.5%, whereas multiple anomalies and unclassified cases represented 13.5% of the study population (Tables 1 and 2).

Table 1: CONGENITAL ANOMALY OF THE SYSTEMS

ANOMALY	1998	1999	2000	2001	2002	2003	2004	2005
GI	10	20	6	3	2	-	4	17
CNS	10	13	9	4	1	2	2	8
CVS	3	4	3	1	-	1	1	2
UGS	-	3	2	-	-	-	2	3
SS	3	6	2	2	-	-	-	3
CT.S	-	2	2	-	-	-	-	-

Table 2: ALL RECOGNISED CONGENITAL ANOMALY

ANOMALY RECOGNISED	FREQUENCY	PERCENTAGE (%)
GENETIC DISORDERS	15	7.5
METABOLIC DISORDERS	3	1.5
GASTROINTESTINAL SYSTEM	61	30.5
CNTRAL NERVOUS SYSTEM	49	24.5
CARDIOVASCULAR SYSTEM	15	7.5
UROGENITAL SYSTEM	10	5.0
SKELETAL SYSTEM	16	8.0
CUTANEOUS SYSTEM	4	2.0
UNCLASSIFIED	27	13.5
<b>TOTAL</b>	<b>200</b>	

SOME TYPES OF CONGENITAL ANOMALIES



**Meningocele**

Courtesy Dr. **Sunday Pam**,  
Consultant neonatologist, JUTH



**Thanatophoric Dwarfism  
with Hydrocephalus**

Courtesy **Dr. Sunday Pam**, Consultant  
neonatologist, JUTH



**Anencephaly**

Courtesy **Dr. Sunday Pam**,  
Consultant neonatologist, JUTH

### 3. DISCUSSION

A total of 200 congenital anomalies were recorded. Congenital anomaly of the gastrointestinal system has the highest occurrence (30.5%), followed by that of the central nervous system with an occurrence of 24.5%. 13.5% represented multiple anomalies and unclassified cases. This includes cases not properly categorised at the point of diagnosis, cases of multiple anomalies that cut across more than one system that could not be grouped alongside those with known genetic/chromosomal abnormalities.

The least occurring anomaly is that of the metabolic disorder with 1.5% occurrence followed by that of the cutaneous system with percentage occurrence of 2.0%. Other congenital anomalies are the urogenital system 5%, Genetic disorders 7.5%, cardiovascular system 7.5% and skeletal system with 8.0% occurrence.

In genetic/chromosomal anomalies, cases of trisomies 13 and 18 i.e., Patau's and Edward's syndromes have the highest occurrence of 26% each, followed by Pierre-Robin's and Down's syndrome with 13.3% occurrence each. Other syndromes include Prune-Belly, Treacher-Collin's and Potter's with 6.7% occurrences each.

In the calculations of the significance difference between the congenital anomaly of the central nervous system and the congenital anomaly of the gastrointestinal system, the calculated  $X^2$  of 1.4 is less than the standard  $X^2$  of 3.84 at a degree of freedom of 1 indicated in the table. Therefore the Null Hypothesis is upheld, meaning that there is no significant difference in the occurrence of the congenital anomalies of the central nervous system and that of the gastrointestinal system.

Omphalocele emerge as the highest occurring congenital anomaly of the gastrointestinal system with 40% occurrence. Imperforate Anus, Gastrointestinal obstruction, Hirschsprung disease and Cleft Palate/lip have occurrence of 25%, 16%, 13.1% and 4.9% respectively.

Myelomeningocele has the highest frequency of occurrence of 57.1% among the congenital anomaly of the central nervous system. Hydrocephalus, encephalocele, meningocele and Neuroblastoma have occurrence of 18.4%, 16.3%, 6.1% and 2% respectively.

Reducing the birth prevalence and associated infant mortality and morbidity attributed to congenital anomalies is an attainable goal. Primary preventive efforts are clearly the optimal approach for ensuring the healthiest possible pregnancy outcomes. Food fortification with folic acid, promoting folic acid-containing multivitamin use in the periconceptional period, pre-pregnancy immunization against rubella, and interventions to reduce alcohol and drug use in pregnancy are examples of important primary preventive efforts.

Prenatal diagnosis and subsequent termination of affected pregnancies, as well as in-utero treatment of prenatally detected congenital anomalies, are two secondary preventive strategies. As the scope of in-utero treatment remains limited, secondary prevention is mainly achieved through selective abortion. Prenatal diagnosis also contributes to tertiary prevention in cases where an early prenatal diagnosis improves postnatal management and reduces or avoids neonatal complications.

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