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Frequency of congenital anomalies in a tertiary health institution in southern Nigeria: A retrospective study from 2016 – 2018

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Abstract

Background/aim: Congenital anomalies continue to be a challenge in neonatal medicine. Improving research and the consequent improvement of prevention and management requires a forensic audit of the frequency of each anomaly in children. Understanding the aetiology of congenital anomaly is important in prevention and in genetic counseling that may help in eradication. Generally, the aetiology of birth defects remains unclear but is thought to be multifactorial.

This retrospective study covering the years 2016 – 2018 was undertaken at the University of Calabar Teaching Hospital (UCTH) in Southern Nigeria. This was a cross sectional retrospective study in which a review of all new born admitted in the New Born Special Baby Care Unit (NBSCU) and also visited the Child Out-patient (CHOP) unit of the University of Calabar Teaching Hospital, Calabar, over a 36 months period (January 2016 - January 2018) was conducted.

Materials and methods: The data was grouped/classified using the 10^{th} Revision of the International Statistical Classification of Diseases and Related Health Problems (ICD-10). The proportions were compared using Chi-square (p < 0.05).

Results: The findings from the 438 cases examined revealed 129 anomalies including congenital malformations of the nervous system, circulatory system, respiratory system, digestive system, genital organs/urinary system, musculoskeletal system, cleft lip and cleft palate and chromosomal abnormalities. *Talipes equinovarus*, congenital malformation of the heart and congenital cataract were the three most prevalent anomalies recorded in the University of Calabar Teaching Hospital. The case files indicated a strong genetic predispositions for these disease conditions. Majority of these anomalies were recorded in outpatients who only visited the hospital for delivery and have missed out on the benefits of managed ante-natal care where some of these defects may have been detected through genetic screenings and scans.

Conclusion: The study concluded there was a significant number of congenital anomalies within the study period.

Key words: Congenital, anomalies, malformations, neonatal, prevalence

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1. Introduction

Congenital abnormality has been a silent catalyst in the increasing rate of neonatal death worldwide. Early intrauterine period (between the 3rd and he 8th week of gestation) is the vital period of life for the normal development of organs, and any injury within that period may result in congenital abnormalities (Malla, 2007). Congenital anomalies, are structural, behavioural, functional and metabolic disorders that occur during intrauterine life and can be identified prenatally, at birth or later in fancy (Malla, 2007; Sadler, 2015). WHO (2016) defined congenital anomalies as defects of structure or function, including metabolism, which can be diagnosed during intrauterine fetal life, at birth, or later in life (WHO, 2015). Most congenital anomalies are life threatening, or can lead to life long and severe disabilities on the neonate.

Generally, the aetiology of birth defects remains unclear but is thought to be multifactorial (Kumar, 2008). For instance, it is known that folate supplementation helps in the prevention of neural tube defects, especially in the first trimester. It is, however, observed that better maternal care an improved standards of living have little effect on the overall frequency of congenital malformations [Hudgins *et al.*, 2008]. Environmental factors such as air pollution and proximity to hazardous waste sites have been recently been reported to increase risk of structural birth defects and chromosomal abnormalities (Vrijheid, 2002; Ritz *et al.*, 2002). Documented rates of congenital anomalies vary from country to country and even in localities within a country. For instance, rates of congenital anomalies noted in Cross River and Akwa Ibom states in the South South region and Kano state in the North-east region of Nigeria are 0.4% and 5.8%, respectively (Mukhtar-Yola *et al.*, 2005; Ekanem *et al.*, 2008). Unfortunately, more than 90% of congenital anomalies occur in low- and middle-income countries (Sitkin *et al.*, 2015). The percentage of the prevalence of congenital anomalies among infants in Iran is 2.3% and is lower than that of some studies conducted in other counties in the world. The rates of congenital anomalies were reported to be 97.6% in Uganda (Ndibazza *et al.*, 2017) and 3.17% in Egypt (Temtamy *et al.*, 1998).

The prevalence rates of congenital anomalies were reported following 2% in nine countries in Latin America (Dolk *et al.*, 2010), 2.7% in Baharan (Sheikha-Salm, 1995), 1.28% and 2.1% in two studies in India (Dutta *et al.*, 2000; Ronya *et al.*, 2002) and 2.0% in Turkey (Oztarhan *et al.*, 2010) which is close to the estimated prevalence rates of anomalies in Iran. However, the prevalence rates of anomalies in Lebanon (1.65%) (Bittarz, 1998) and in China (1.5%) (Cheng, 2003) are lower than the estimations in Iran. The Difference in the prevalence rates of congenital anomalies in different parts of the world can be due to differences in genetics, racial, cultural and socio-economic factors among individuals and the assessment method of infants (Vantankhan *et al.*, 2016).

Most developed countries have congenital anomalies registry, and epidemiological surveillance of malformations with potential teratogenic environmental agents (Matthews *et al.*, 2001; Lowry *et al.*, 2002). The reported incidence and mortality rates associated with congenital anomalies remained speculatively high, with very little information on other epidemiological data.

According to the study carried out in central part of Cross River state, Nigeria from 1992 – 2005, the major anomalies observed were anomalies associated with central nervous system, cardiovascular system, gastrointestinal tract and skeletal system. The highest occurred anomalies were hydrocephalus and tracheoesophageal (Eluwa *et al.*, 2013). Some of the most occurring anomalies include; Down Syndrome, Hydrocephalus, Cerebral Palsy, *Spina Bifida*, *Tracheoesophageal Fistula*, Cleft Lip and Palate, Ventricular septal defect, Polycystic kidney disease, Omphalocele, *Talipes equinovarus*, and *Encephalocele*.

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2. Materials and methods

This was a cross sectional retrospective study in which a review of all new born admitted in the New Born Special Baby Care Unit (NBSCU) and also visited the Children Out-patient (CHOP) unit of the University of Calabar Teaching Hospital, Calabar, over a 36 months period (January 2016 - January 2018). The case files of these babies were retrieved from the hospital records unit and examined individually to identify the type of anomaly that was recorded. Ethical approval was sought and obtained from the Ethical Committee of the University of Calabar Teaching Hospital.

The data used for this study was gotten from only neonates who have been diagnosed with any congenital anomaly. Neonates without any case or record of an anomaly were totally excluded from this research.

Data analysis

The Data was grouped/classified using the 10^{th} Revision of the International Statistical Classification of Diseases and Related Health Problems (ICD-10). Rates and proportions were calculated with 95% confidence intervals. The proportions were compared using Chi-square with the level of significance set at P < 0.05.

3. Results

A total of four hundred and thirty-eight (438) babies were observed to have one hundred and twenty-nine (129) different anomalies, between January 2016- December 2018, in the University of Calabar Teaching Hospital (UCTH), Calabar.

The anomalies were grouped and classified using the International Statistical Classification of Diseases and related health problems (ICD-10). Table 1 shows the frequency and percentage of these anomalies as classified using ICD-10. The anomalies were classified into groups which are; Congenital malformation of the nervous system, Congenital malformation of the eye, ear face and neck, Congenital malformation of the circulatory system, Congenital malformation of respiratory system, Cleft Lip and Cleft Palate, Other congenital malformation of the digestive system, Congenital malformations of genital organs, Congenital malformation of genital organs, Congenital malformations and deformities of the musculoskeletal system, Other congenital malformations, and Chromosomal abnormalities, not elsewhere specified.

Figure 1 to Figure 11 shows the bar chart of the individual anomaly in their respective group, showing the most common i.e. one with the highest frequency. At 95% confidence interval, there was significance in the observed frequency (p<0.05).

4. Discussion

Nigeria ranks only second to India in the absolute number of annual neonatal deaths worldwide (Ezeh *et al.*, 2014; Akinyemi *et al.*, 2015). Congenital anomalies contribute significantly to this burden, particularly in developing countries (Sachdeva *et al.*, 2014).

This study was a three-year review of the frequency of congenital anomalies in the University of Calabar Teaching Hospital (UCTH), Calabar. An annual increase in the frequency of these anomalies where observed that is, in 2016, 109 babies (24.9%) were congenitally malformed, in 2017, 151 babies (34.5%) were congenitally malformed, and in 2018, 178 babies (40.6%) were recorded to be congenitally malformed. It was observed that every year, the higher frequencies of these anomalies were recorded from outpatient rather than inpatients. This therefore coincide with the work of Bukar and Jauro (2013) in the north-east that noted that about 90% of women who had children with birth defects did not attend ante-natal clinic but preferred to deliver their babies outside the teaching hospital with traditional birth attendants in attendance.

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Current study reported congenital malformations and deformities of the musculoskeletal system as the most common anomaly in our study which can be compared with the study of congenital malformation done at a referral hospital in Gorgan, Islamic Republic of Iran in 1998-1999, where anomalies of musculoskeletal system had the highest incidence. Onyearugha *et al.* (2014) observed that the highest prevalence of congenital anomalies was recorded in the digestive system. This is similar to the observations in studies conducted in Abia state University Teaching Hospital Aba, Abia state which is the same geopolitical zone and the highest recorded in the gastrointestinal tract (Ekanem *et al.*, 2004). In contrast, central nervous system anomalies were the most frequent congenital anomalies recorded in studies carried out in Akwa-Ibom, Rivers and Lagos states of Nigeria. The difference between the between the body systems manifesting most frequent malformations in the different locations of the country aforementioned is not readily explainable. Some other authors have reported predominance of anomalies of the skeletal and nervous system having focused mainly on externally visible anomalies or self-report from population-base study.

It is generally known that maternal lifestyle may predispose to the development of congenital anomaly in their offspring. The consumption of alcohol, cigarette and certain medication are known teratogenic factors (Sadler *et al.*, 2015). Pollution from the petrochemical industries has been reported as causing birth defects (Oliveira *et al.*, 2002). Toxic agents might affect particularly the development of the central nervous system which occurs in the 4th and 5th weeks of gestation. Lack of good and affordable health care encourages pregnant women to resort patronage of traditional birth attendants who give them herbal drugs and concoction at unknown doses. This certainly may affect the developing fetuses. Some of these resort to self-medication and unprescribed drugs especially anti-malaria drugs. Many do not predispose themselves to antenatal clinics where they could have been advised against indiscriminate use of drugs during pregnancy (Rottlaender *et al.*, 2008). Moreover, evidence has shown association with some genetic determinants (such as mutations in folate-responsive or folate-dependent pathways) which could affect the central nervous system development adversely from conception (Kinsman *et al.*, 2011).

This study reports Talipes equinovarus as the most common anomaly in UCTH. Talipes equinovarus is a common foot abnormality also known as club foot, in which the foot points downward and inward. According to Lloyd Roberts, Talipes equinovarus is the commonest congenital foot abnormality and remains the most difficult to treat. In 1964, Wynne-Davies suggested that there is a mixed genetic and environmental causes of Talipes equinovarus. From his study, if one child in the family has the deformity, the chances of the second having it is 20 times that in the general population. This therefore suggests that, Talipes equinovarus is highly inheritable. Intrauterine environmental causes have been suggested because of the similarity of the deformity with that found in weakness of peroneal muscles of neurological originor contracture of the plantar flexor and inverter muscles. While in 1936, Browne suggested the cause to be due to increased intrauterine fluid tension. Based on current study, talipes equinovarus was found mostly among females than males, which suggests that it may or may not be sex linked.

Congenital cataract which is the second most common anomaly in The University of Calabar Teaching Hospital (UCTH), is an opacification of the eye lens appearing at birth or shortly thereafter. Inherited eye diseases comprise 1/3 or all reported human genetic disorders (Khan *et al.*, 2018). This anomaly is one of the highest significant causes of blindness or optical impairment in the childhood. A significant amount of congenital cataract has monogenic roots, it is genetically heterogeneous, and all the modes of Mendelian inheritance have been described. Numerous genes linked with inherited and pediatric cataracts have been recognized. Inherited congenial cataract has been linked with mutations in particular genes, comprising of crystallins, membrane transport channel proteins, gap junction proteins, the development transcriptional factors and the cytoskeleton (Khan *et al.*, 2008).

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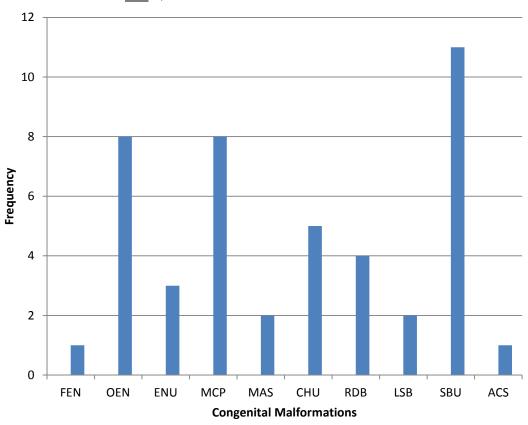
However, we may have significantly under-estimated the actual incidence of the anomalies in the general population. The study area being a tertiary health facility, complicated cases are more frequently seen, while the uncomplicated cases may have been treated in other peripheral facilities.

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Table 1: Frequency and Percentage of Congenital Anomalies recorded in UCTH from January 2016 – December 2018 as classified using ICD-10

	S	Malformation	Frequen	Percenta
/N			су	ge
	1	Congenital Malformations of the Nervous system	45	10.3%
	2	Congenital Malformation of eye, ear, face, and neck	63	14.5%
	3	Congenital Malformation of the Circulatory System	42	9.6%
	4	Congenital Malformation of Respiratory System	7	1.6%
	5	Cleft Lip & Cleft Palate	8	1.8%
	6	Other Congenital Malformation of the Digestive System	27	6.3%
	7	Other Congenital Malformation of Genital organs	48	11%
	8	Congenital Malformation of Urinary System	30	6.8%
	9	Congenital Malformation and Deformities of the	125	28.5%
		Musculoskeletal System		
	1	Other Congenital Malformations	22	5.0%
0				
	1	Chromosomal Abnormalities, not elsewhere classified	21	4.8%
1				





Key

- FEN- Frontal Encephalocele
 OEN- Occipital Encephalocele
 ENU- Encephalocele, Unspecified
- MCP- Microcephaly
- MAS- Malformation of Aqueduct of Sylvius CHU- Congenital Hydrocephalus, Unspecified RDB- Other Reduction Deformities of Brain
- LSB- Lumbar Spina Bifida without
 - Hydrocephalus
- SBU- Spina Bifida, Unspecified ACS- Arnold-chiari Syndrome

Figure 1: Bar chart showing the frequency of Congenital Malformation of the Nervous System from January 2016- December 2018

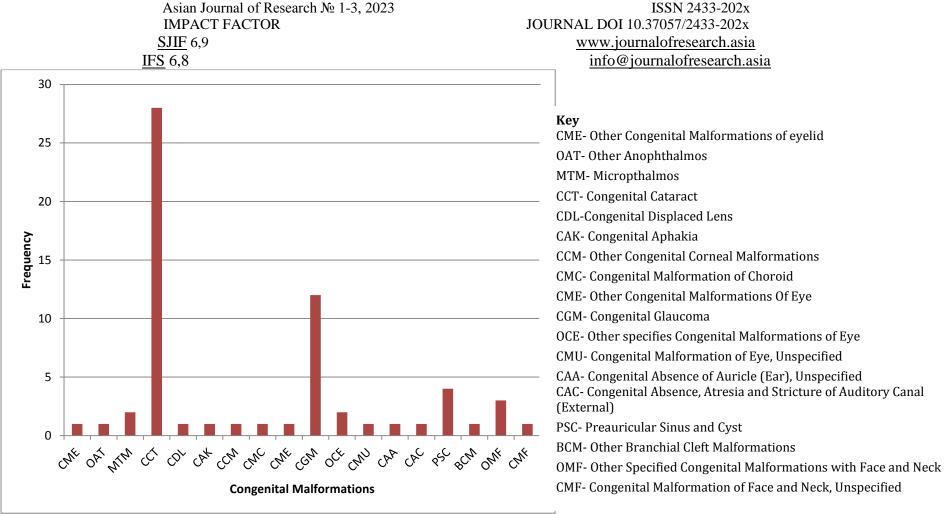
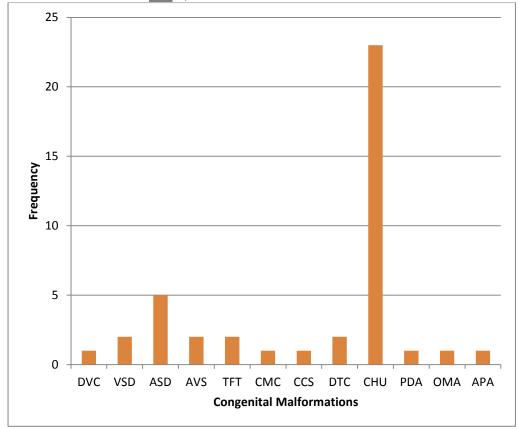


Figure 2: Bar chart showing the frequency of Congenital Malformation of Circulatory System from January 2016- December 2018



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Key

- DVC-Discordant Ventriculoarterial Connection
- VSD- Ventricular Septal Defect
- ASD- Atrial Septal Defect
- AVS- Atrioventricular Septal Defect
- TFT- Tetralogy of Fallot
- CMC- Other Congenital Malformation Of Cardiac Septa
- CCS- Congenital Malformation of Cardiac Septum, Unspecified
- DTC- Dextrocardia
- CHU- Congenital Malformation Of Heart, Unspecified
- PDA- Patent DuctusArteriosus
- OMA- Other Congenital Malformations Of Aorta
- APA- Atresia of Pulmonary Artery

Figure 3: Bar chart Showing the frequency of Congenital Malformation of Circulatory System from January 2016- December 2018

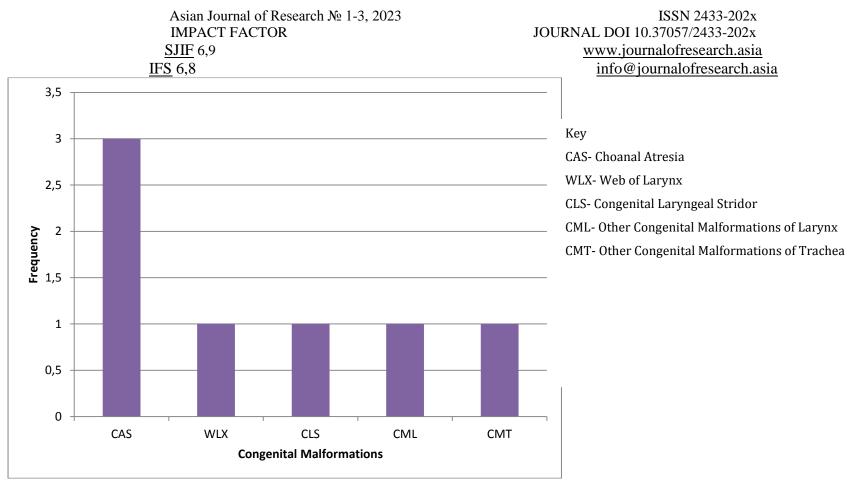
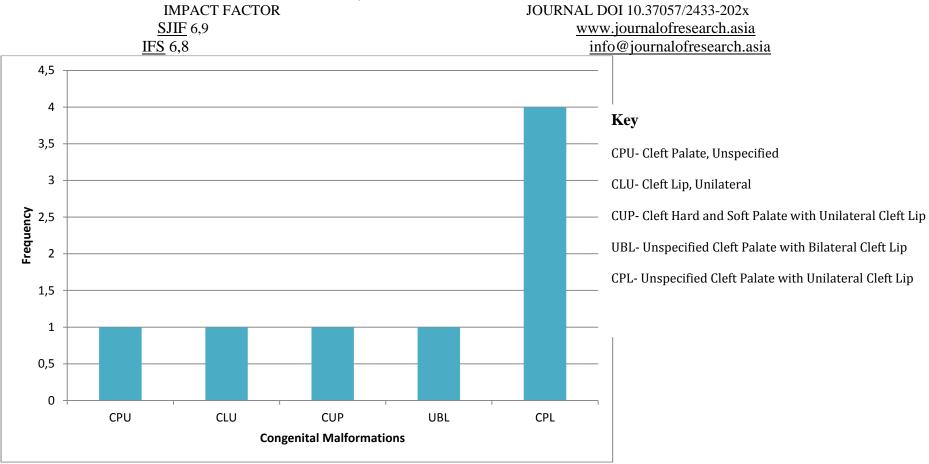


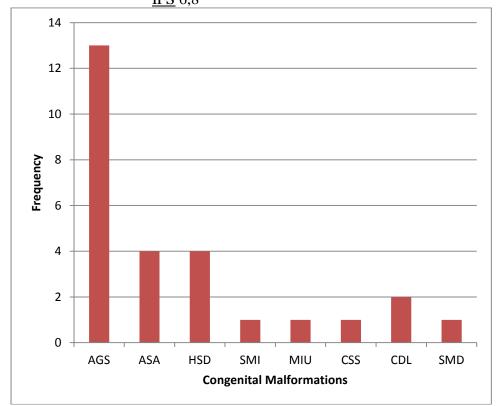
Figure 4: Bar chart showing the frequency of Congenital Malformation of the Respiratory System from January 2016- December 2018



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Figure 5: Bar chart showing the frequency of Cleft Lip & Cleft Palate from January 2016- December 2018

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Key

AGS- Ankyloglossia

ASA- Congenital absence, Atresia and Stenosis Of Anus without Fistula

HSD- Hirschsprung's Disease

SMI- Other Specified Congenital Malformation of Intestine

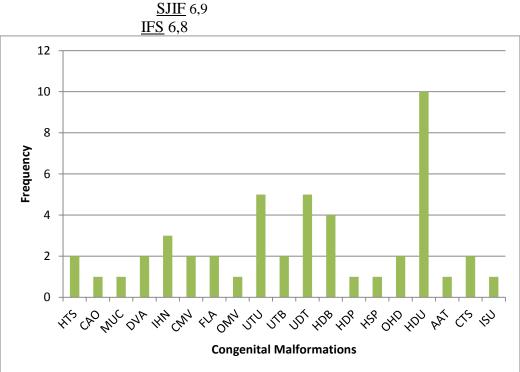
MIU- Congenital Malformation of Intestine, Unspecified

CSS- Congenital Stenosis and Stricture of Bile Ducts

CDL- Cystic Disease of Liver

SMD- Other Specified Congenital Malformations of Digestive System

Figure 6: Bar chart showing the frequency of Congenital Malformation of Digestive system from January 2016- December 2018



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IMPACT FACTOR

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Key

THE- Hypoplasia Of Testis and Scrotum

CAO- Congenital Absence of Ovary

MUC- Other Congenital Malformation of Uterus and Cervix

DVA- Doubling of Vagina IHN- Imperforate Hymen

CMV- Other Congenital Malformation of Vagina

FLA- Fusion of Labia

OMV- Other Congenital Malformation of Vulva

UTU- Undescended Testicle, Unilateral

UTB- Undescended Testicle, Bilateral

UDT- Undescended Testicle, Unspecified

HDB- Hypospadias, Balanic

HDP- Hypospadias, Penile

HSP- Hypospadias, Penoscrotal

OHD- Other Hypospadias

HDU- Hypospadias, Unspecified

AAT- Absence of Aplasia of Testis

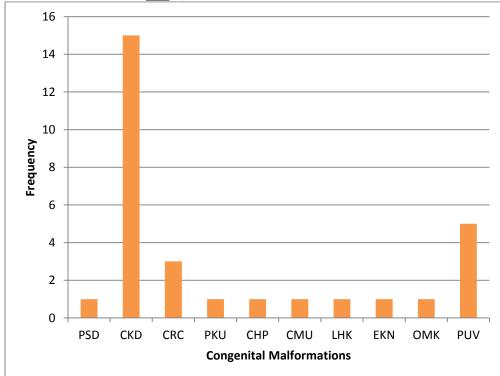
CTS- Other Congenital Malformations of Testis and scrotum

ISU- Indeterminate Sex, Unspecified

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Figure 7: Bar chart showing the frequency of Congenital Malformation of Genital Organs from January 2016- December 2018





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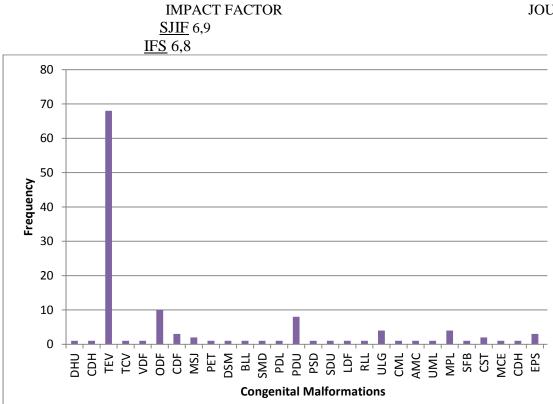
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Key

- PSD- Potter's Syndrome
- CKD- Cystic Kidney Disease
- CRC- Congenital Single Renal Cyst
- PKU- Polycystic Kidney, unspecified
- CHP- Congenital Hydronephrosis
- CMU- Other Congenital Malformation Of Ureter
- LHK- Lobulated, Fused and Horeseshoe Kidney
- **EKN-** Ectopic Kidney
- OMK- Other Specified Congenital Malformation of Kidney
- PUV- Congenital Posterior Urethral Valves

Figure 8: Bar chart showing the frequency of Congenital Malformation of Urinary System from January 2016- December 2018



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Figure 9: Bar chart showing the frequency of Congenital Malformation and Deformit January 2016- December 2018

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Key
DHU- Congenital Dislocation of hip, Unilateral
CDH- Other Congenital Deformities of Hip
TEV- TalipesEquinovarus
TCV- TalipesCalcaneovarous
VDF- Other Congenital Valgus Deformities of Feet
ODF- Other Congenital Deformities of Feet
CDF- Congenital Deformity of Feet, Unspecified
MSJ- Other Congenital Malformation of Skull, Face, & Jaw
PET- PectusExcavatum
DSM- Congenital Deformity of Sternocleidomastoid Muscle
BLL- Congenital Bowing of Long Bones of leg, Unspecified
SMD- Other Specified Congenital Muscoloskeletal Deformities
PDL- Polydactyly
PDU- Polydactyly, Unspecified
PSD- Polysyndactyly
SDU- Syndactyly, Unspecified
LDF- Longitudinal Reduction Defect of Femur
RLL- Other reduction of Lower Limb(s)
ULG- Other Congenital Malformation of the Upper Limb(s), including Shoulder Girdle
CML- Other Congenital Malformation of Lower Limb(s)
AMC- Arthogryposis Multiplex Congenita
UML- Unspecified Congenital Malformation of Limb(s)
MPL- Macrophaly

CST- Congenital Spondylolisthesis

SFB- Other Congenital Malformation of Skull and Face Bones

GTC- Gastroschisis

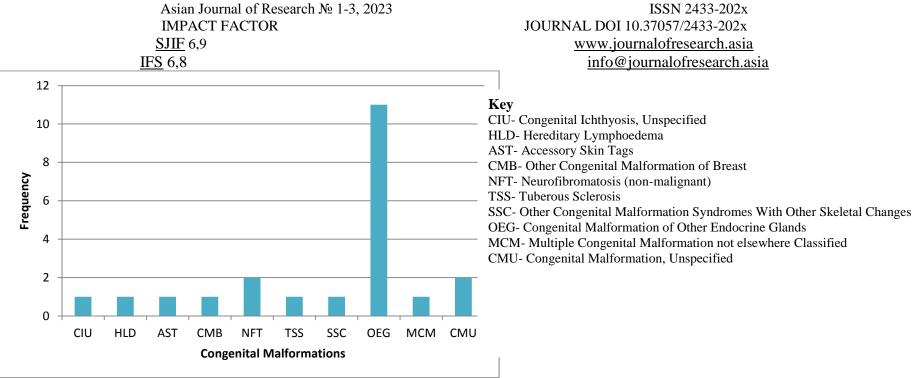


Figure 10: Bar chart showing the frequency of Other Congenital Malformation from January 2016- December 2018

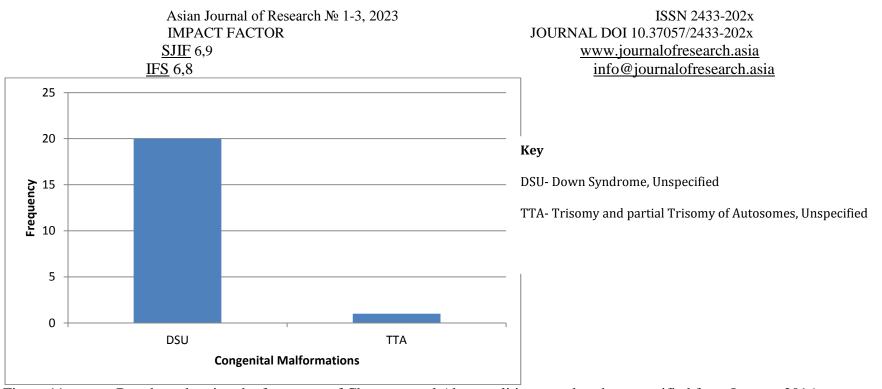


Figure 11: Bar chart showing the frequency of Chromosomal Abnormalities, not elsewhere specified from January 2016-December 2018

Ethical Approval

Ethical approval was sought and obtained from the University of Calabar Teaching Hospital Health Research Ethics Committee (UCTH HREC). The approval number was UCTH NHREC REG NUMBER: NHREC07/10/2012 and HREC Protocol Assigned Number: UCTH/HREC/33/684.

Conflict of interest

The authors declare no competing interest.

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