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Congenital Anomalies at a Nigerian Teaching Hospital: A Retrospective Study, 2016–2018

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Abstract

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. This retrospective study covering the years 2016–2018 was undertaken at the University of Calabar Teaching Hospital (UCTH) in Southern Nigeria. 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 UCTH. The case files indicated a strong genetic predisposition for these conditions. The majority of anomalies were recorded in outpatients who only visited the hospital for delivery and had missed the benefits of managed antenatal care, where some defects may have been detected through genetic screening and ultrasound scans. The study concluded that there was a significant number of congenital anomalies within the study period.

Keywords: Congenital; anomalies; malformations; neonatal; prevalence

Abbreviations: UCTH: University of Calabar Teaching Hospital; WHO: World Health Organization; CHOP: Children Outpatient Unit; ICD-10: International Statistical Classification of Diseases and Related Health Problems, 10th Revision; NBSCU: New Born Special Baby Care Unit

Introduction

Congenital abnormalities have been a silent catalyst in the increasing rate of neonatal death worldwide. The early intrauterine period (between the 3rd and the 8th week of gestation) is the vital period of life for the normal development of organs [1], and any insult within that period may result in congenital abnormalities. Congenital anomalies are structural, behavioural, functional, and metabolic disorders that occur during intrauterine life and can be identified prenatally, at birth, or later in infancy [2, 3]. WHO defines congenital anomalies as defects of structure or function, including metabolism, which can be diagnosed during intrauterine fetal life,

at birth, or later in life [3]. Most congenital anomalies are life-threatening or can lead to lifelong and severe disabilities in the neonate.

Generally, the aetiology of birth defects remains unclear but is thought to be multifactorial [4]. For instance, folate supplementation helps prevent neural tube defects, especially in the first trimester. Environmental factors such as air pollution and proximity to hazardous waste sites have been reported to increase the risk of structural birth defects and chromosomal abnormalities [5, 6]. Documented rates of congenital anomalies vary from country to country and even in localities within a country. For instance, rates of congenital anomalies in Cross River and Akwa Ibom states in the South region, and in Kano state in the North-east region of Nigeria are 0.4% and 5.8%, respectively [7, 8]. Unfortunately, more than 90% of congenital anomalies occur in low- and middle-income countries [9]. The prevalence of congenital anomalies among infants was reported as 97.6% in Uganda [10] and 3.17% in Egypt [11].

The prevalence rates of congenital anomalies were reported as approximately 2% in nine Latin American countries [12], 2.7% in Bahrain [13], 1.28% and 2.1% in two studies in India [14, 15], and 2.0% in Turkey [16]. The prevalence rates in Lebanon (1.65%) [17] and China (1.5%) [18] are comparatively lower. Differences in prevalence rates across regions may be attributed to genetic differences, racial, cultural, and socio-economic factors, as well as the assessment methods used [19].

Most developed countries have congenital anomalies registries and epidemiological surveillance systems for malformations with potential teratogenic environmental agents [20]. The reported incidence and mortality rates associated with congenital anomalies remain speculatively high, with very little information on other epidemiological data.

According to a study carried out in the central part of Cross River State, Nigeria, from 1992 to 2005, the major anomalies observed were associated with the central nervous system, cardiovascular system, gastrointestinal tract, and skeletal system. The highest occurring anomalies were hydrocephalus and tracheoesophageal fistula [21]. Some of the most commonly 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. This study aimed to assess the frequency of congenital anomalies at the University of Calabar Teaching Hospital (UCTH) over a three-year period (2016–2018).

Methods

This was a cross-sectional retrospective study in which a review of all newborns admitted to the Newborn Special Baby Care Unit (NBSCU) and those who visited the Children Outpatient Unit (CHOP) of the University of Calabar Teaching Hospital, Calabar, over a 36-month period (January 2016 – December 2018) was conducted. The case files of these babies were retrieved from the hospital records unit and examined individually to identify the type of anomaly recorded. Ethical approval was sought and obtained from the Ethical Committee of the University of Calabar Teaching Hospital.

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

Data analysis

Data were grouped and classified using the 10th Revision of the International Statistical Classification of Diseases and Related Health Problems (ICD-10). Rates and proportions were calculated with 95% confidence intervals. Proportions were compared using the chi-square test, with a significance level of $P < 0.05$.

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 and December 2018 at 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 the following groups: congenital malformation of the nervous system; congenital malformation of the eye, ear, face, and neck; congenital malformation of the circulatory system; congenital malformation of the respiratory system; cleft lip and cleft palate; other congenital malformation of the digestive system; congenital malformations of genital organs; congenital

malformations and deformities of the musculoskeletal system; other congenital malformations; and chromosomal abnormalities, not elsewhere specified.

Figures 1 to 11 show bar charts of the individual anomalies in their respective groups, illustrating the most common anomaly (i.e., the one with the highest frequency) within each group. At a 95% confidence interval, there was significance in the observed frequency ($P < 0.05$).

Table 1 Frequency and percentage of congenital anomalies recorded in UCTH from January 2016 to December 2018, classified by ICD-10

S/N	Malformation	Frequency	Percentage (%)
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 the respiratory system	7	1.6
5	Cleft lip and cleft palate	8	1.8
6	Other congenital malformation of the	27	6.3

	digestive system		
7	Other congenital malformation of genital organs	48	11.0
8	Congenital malformation of the urinary system	30	6.8
9	Congenital malformation and deformities of the musculoskeletal system	125	28.5
10	Other congenital malformations	22	5.0
11	Chromosomal abnormalities, not elsewhere classified	21	4.8
Total		438	100.0

Figure captions

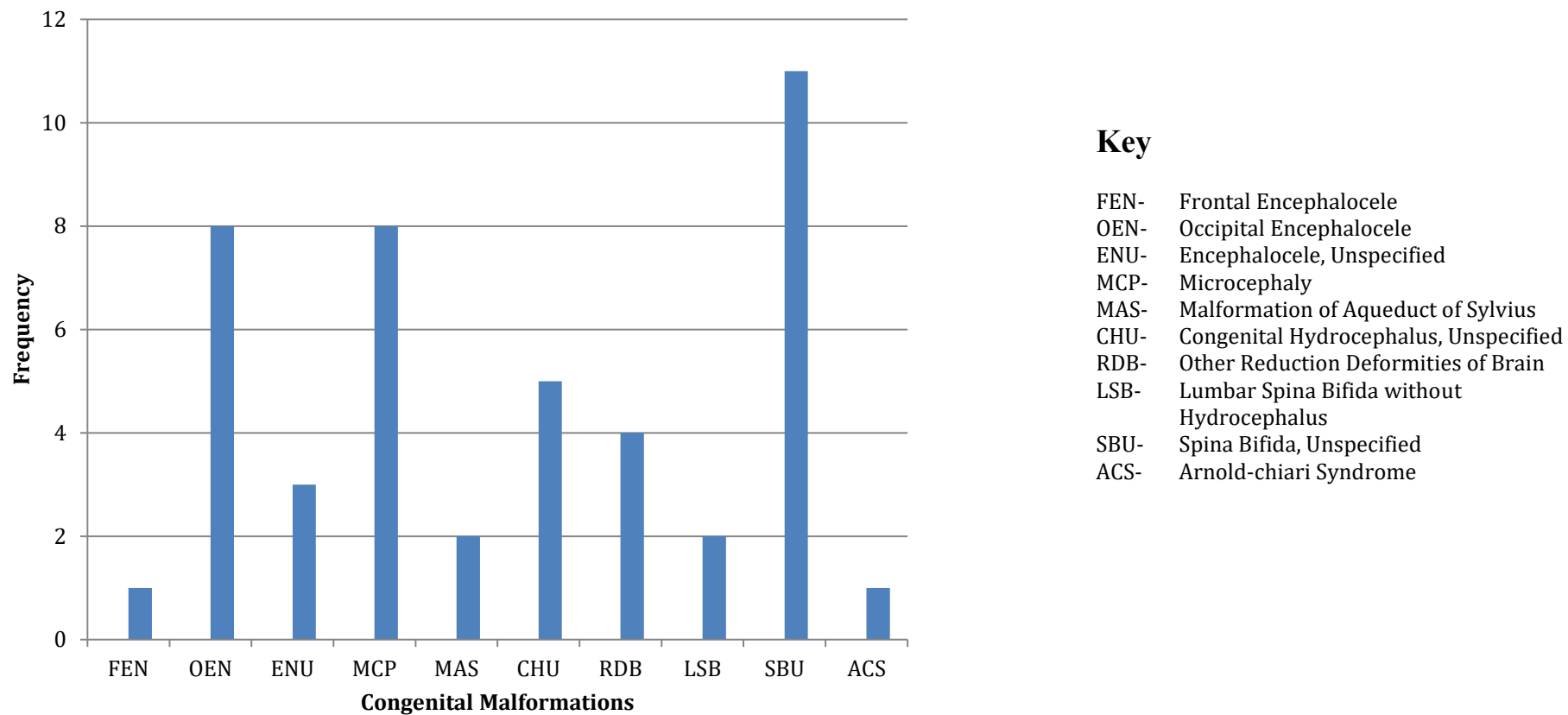


Fig. 1 Bar chart showing the frequency of congenital malformations of the nervous system, January 2016–December 2018

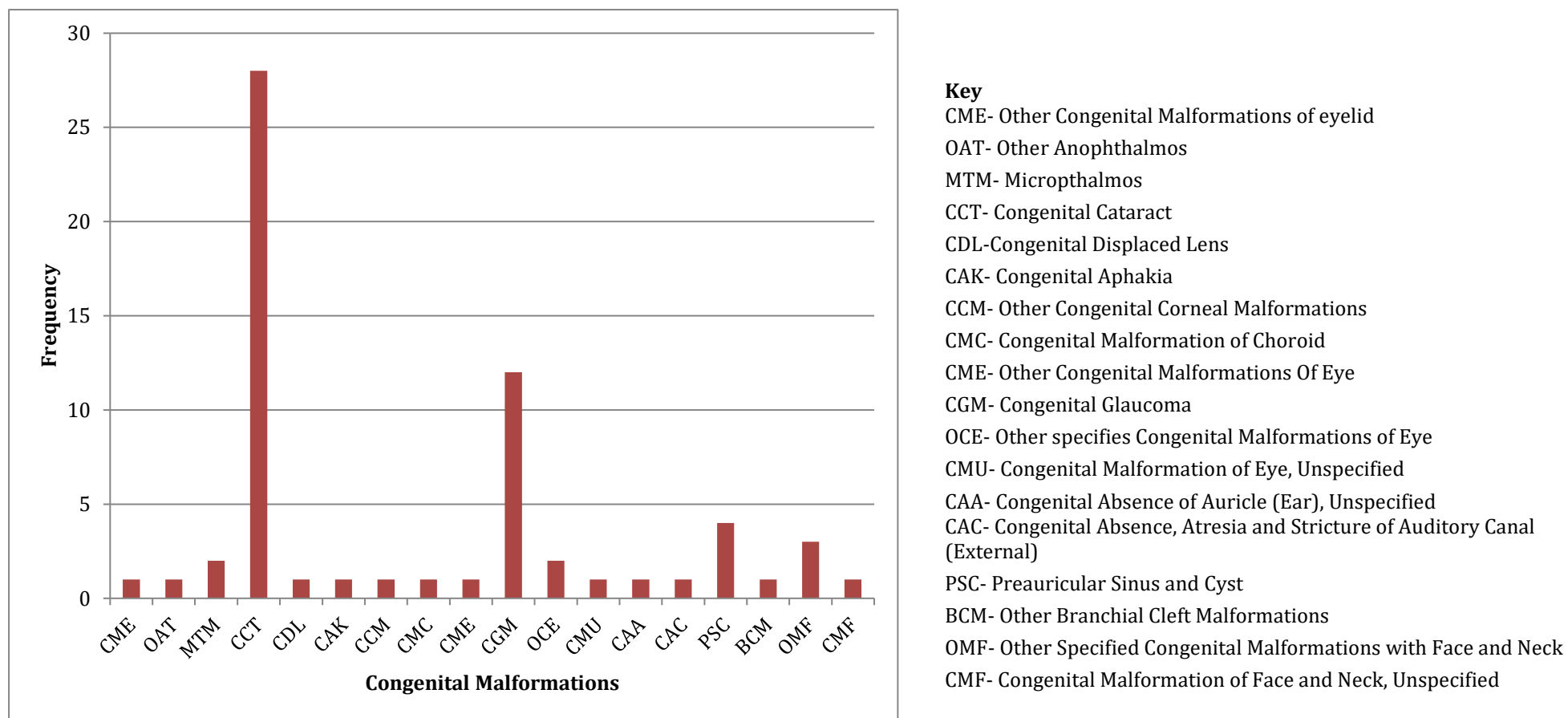


Fig. 2 Bar chart showing the frequency of congenital malformations of the eye, ear, face, and neck, January 2016–December 2018

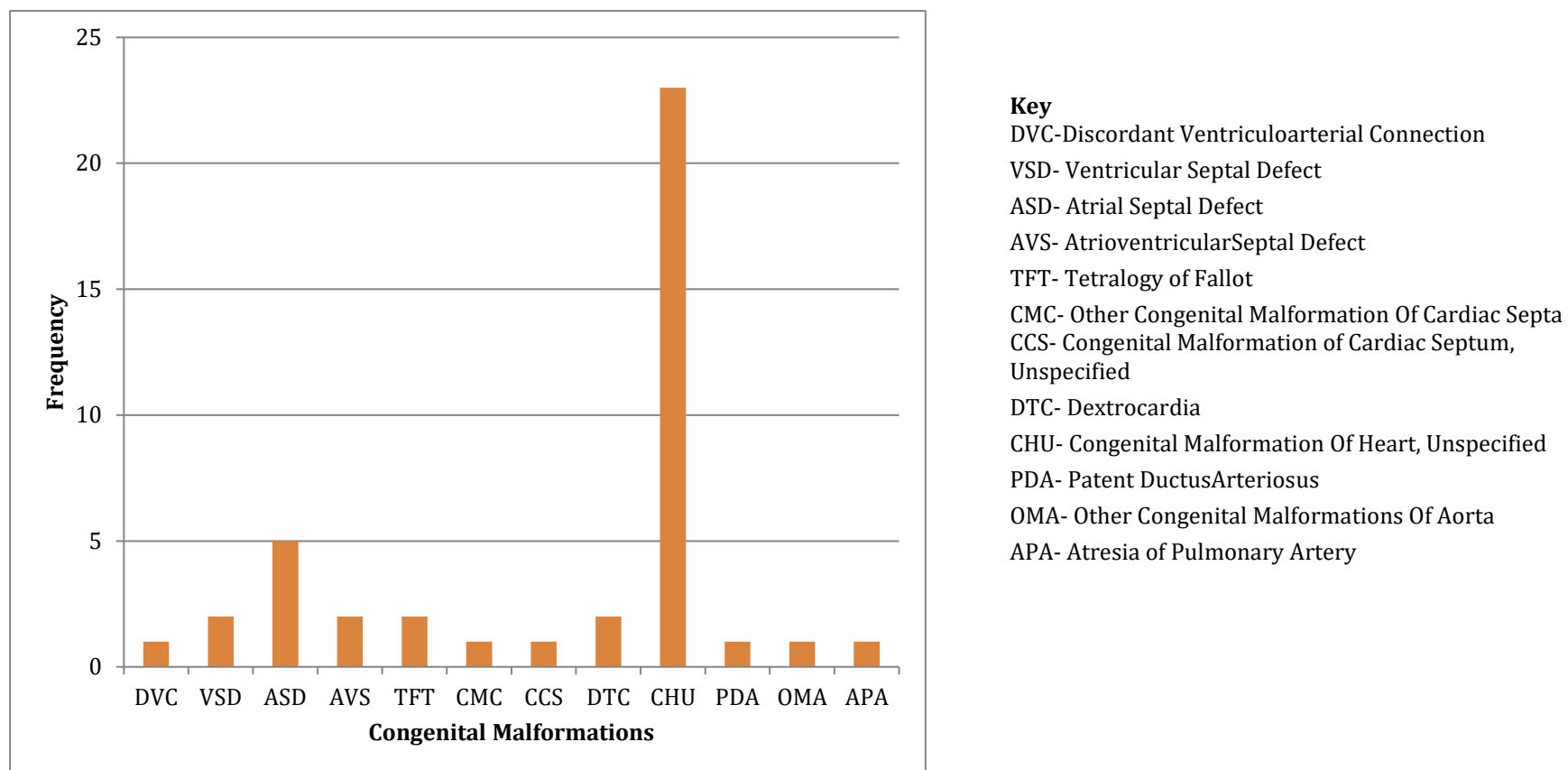


Fig. 3 Bar chart showing the frequency of congenital malformations of the circulatory system, January 2016–December 2018

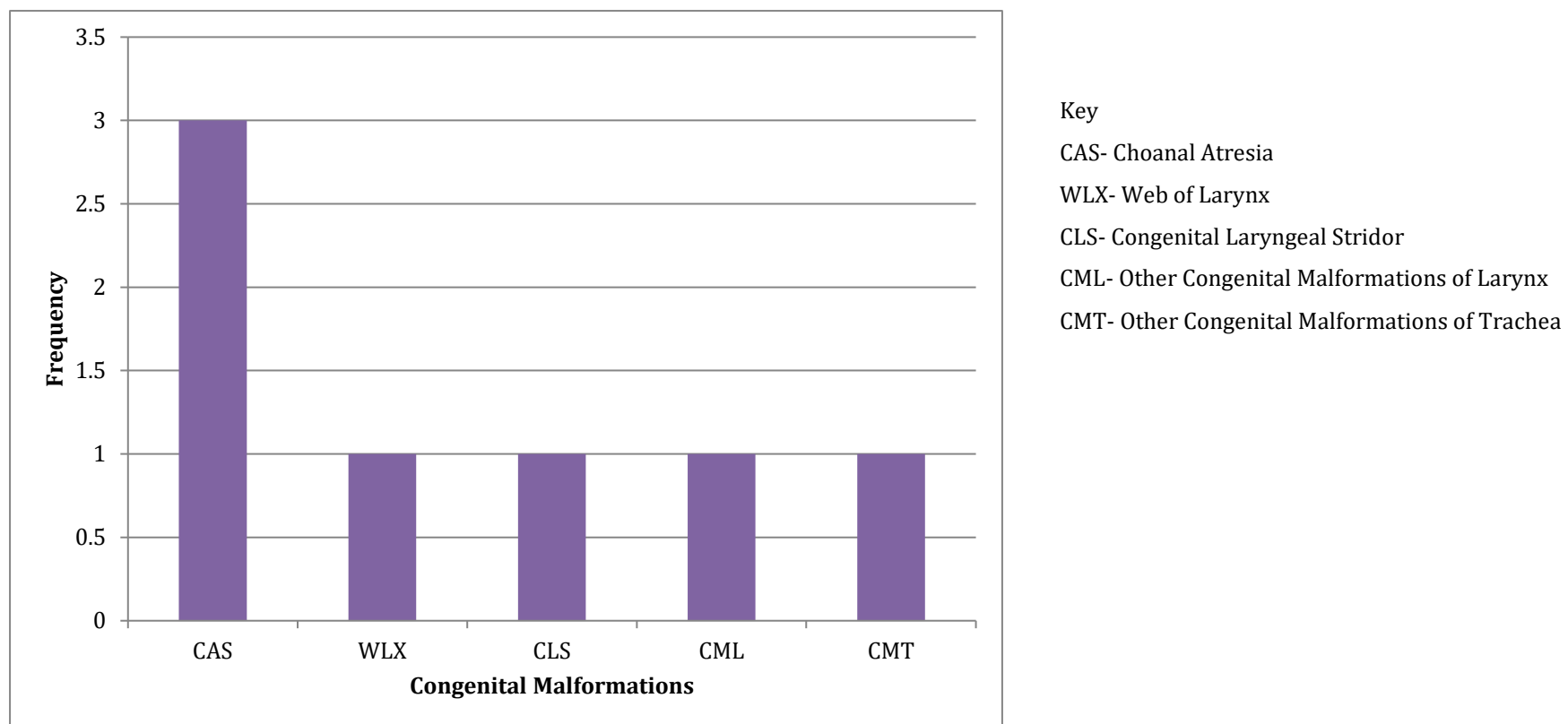


Fig. 4 Bar chart showing the frequency of congenital malformations of the respiratory system, January 2016–December 2018

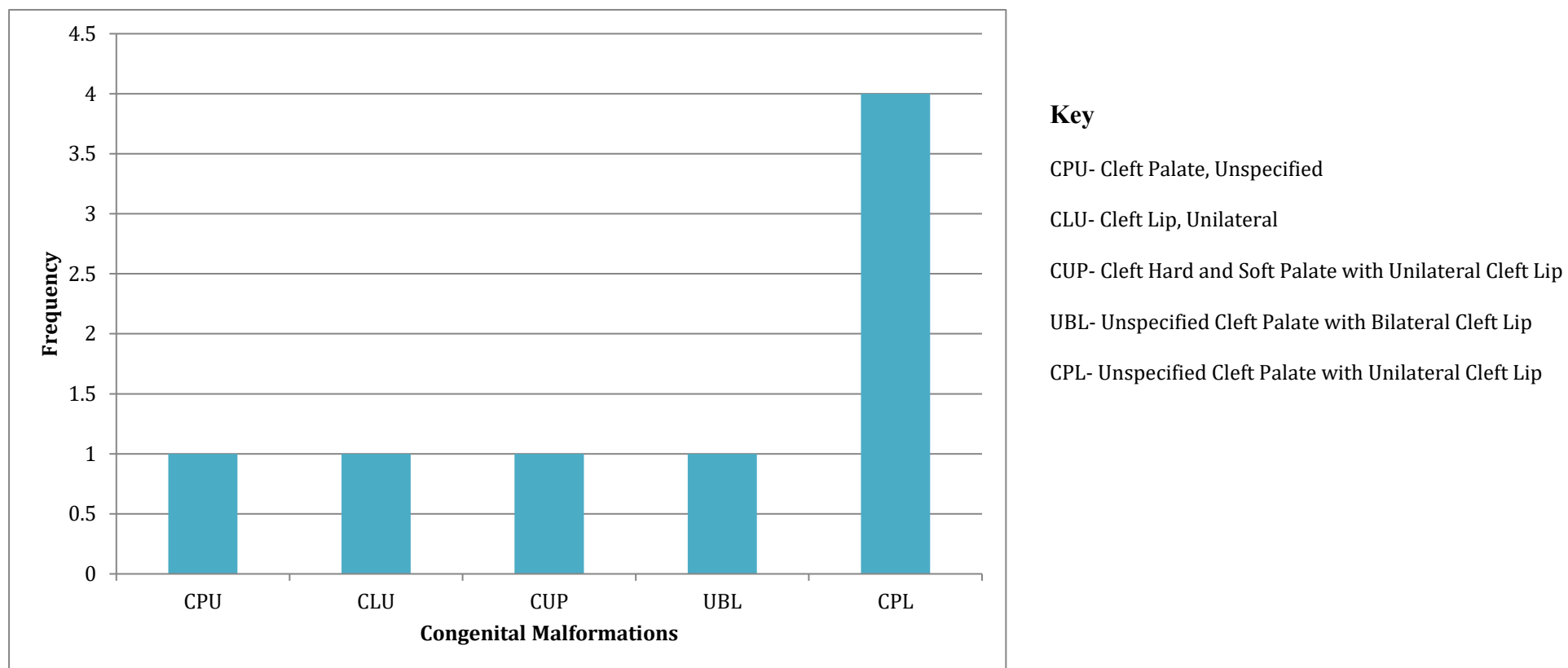
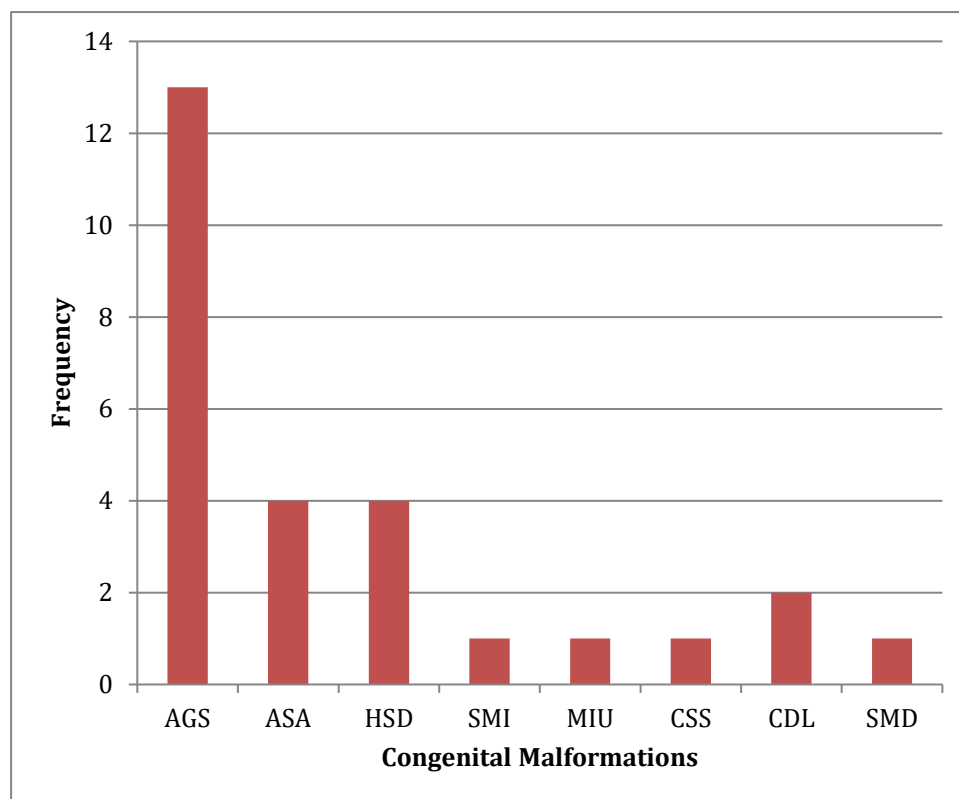


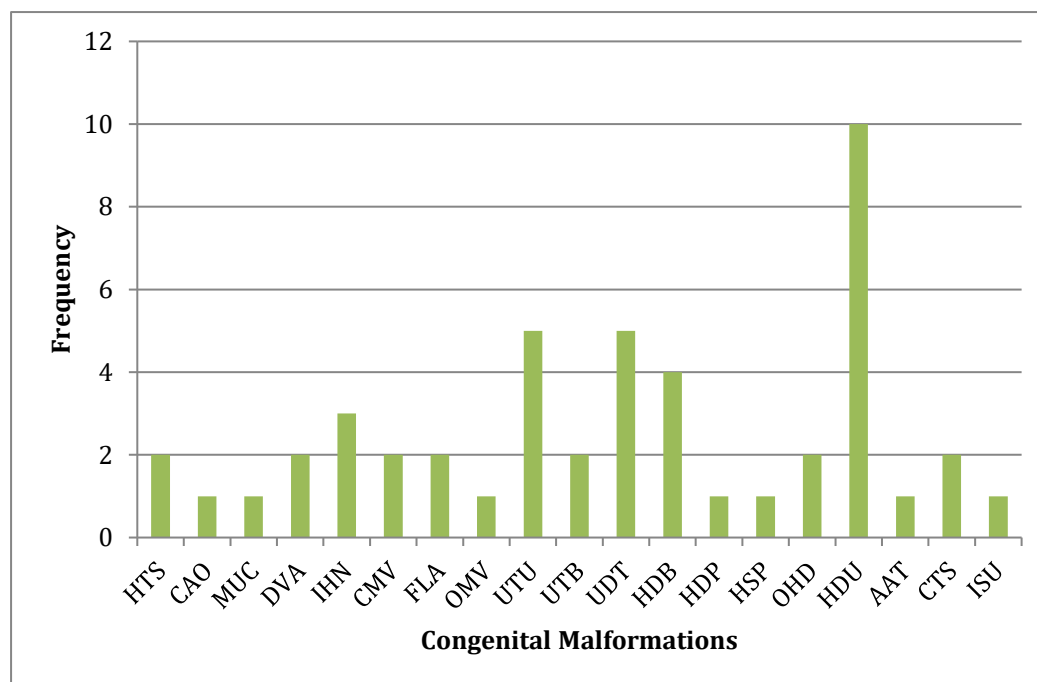
Fig. 5 Bar chart showing the frequency of cleft lip and cleft palate, January 2016–December 2018



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

Fig. 6 Bar chart showing the frequency of congenital malformations of the digestive system, January 2016–December 2018



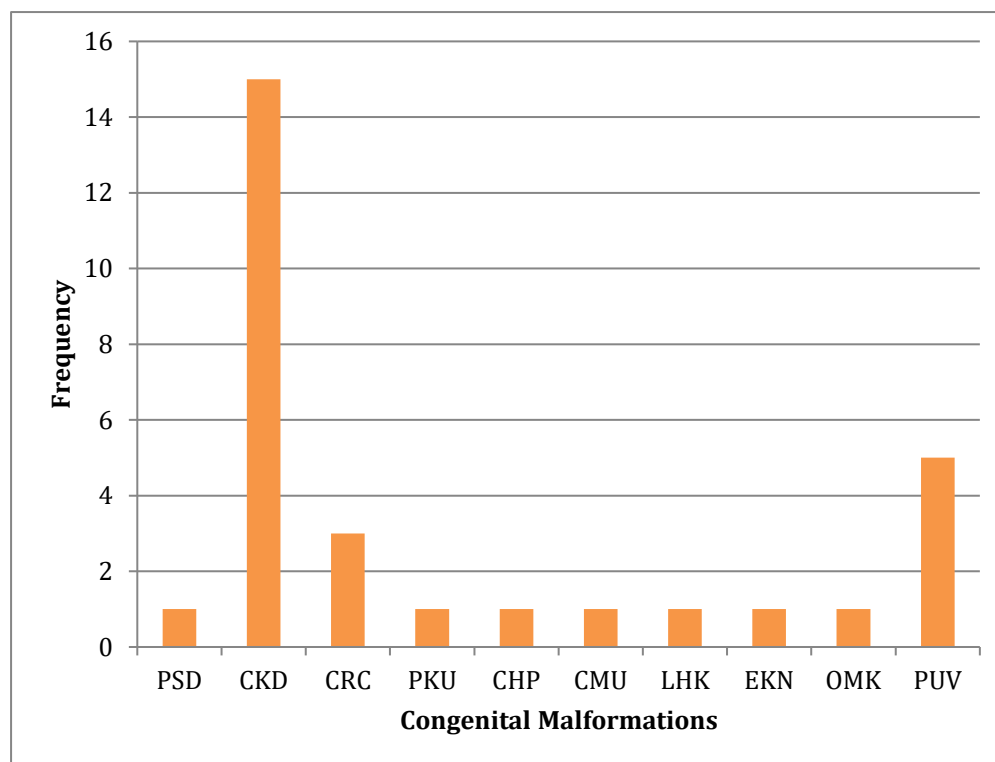
Key

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

6

7 Fig. 7 Bar chart showing the frequency of congenital malformations of genital organs, January 2016–December 2018

8



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

9

10 Fig. 8 Bar chart showing the frequency of congenital malformations of the urinary system, January 2016–December 2018

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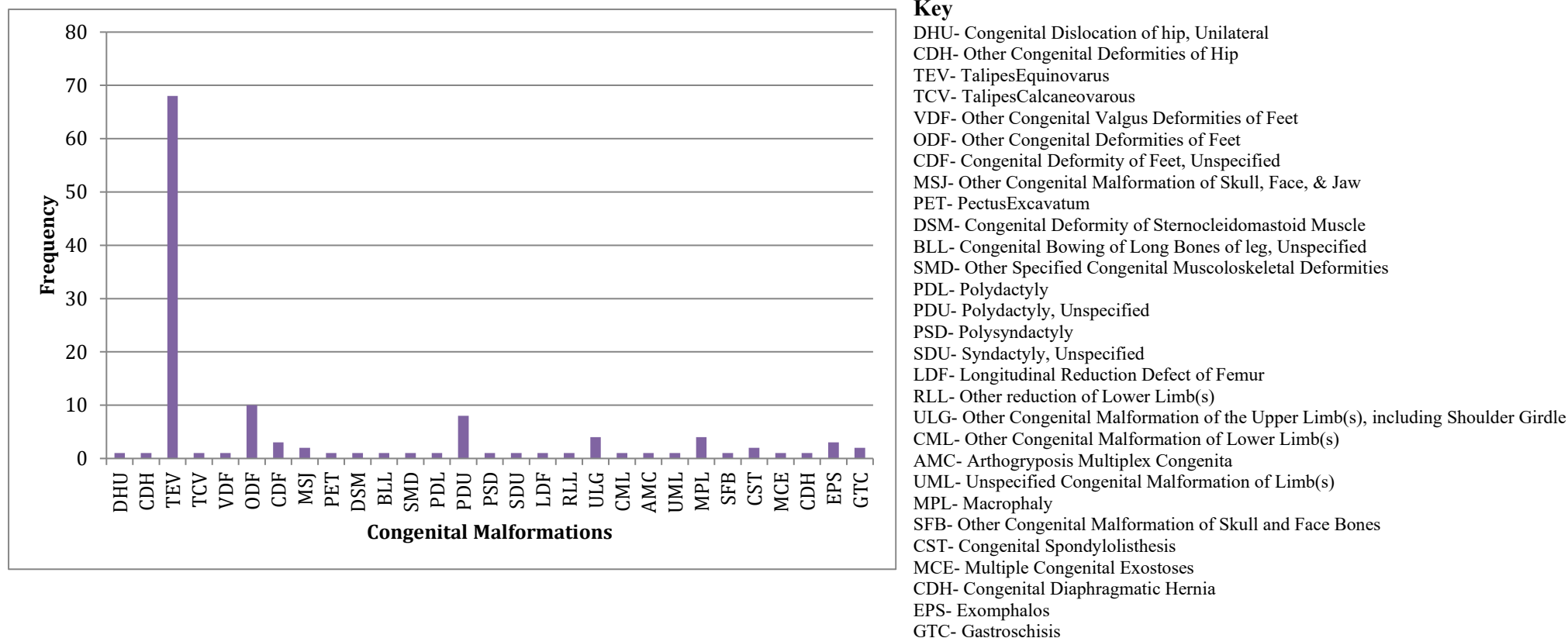
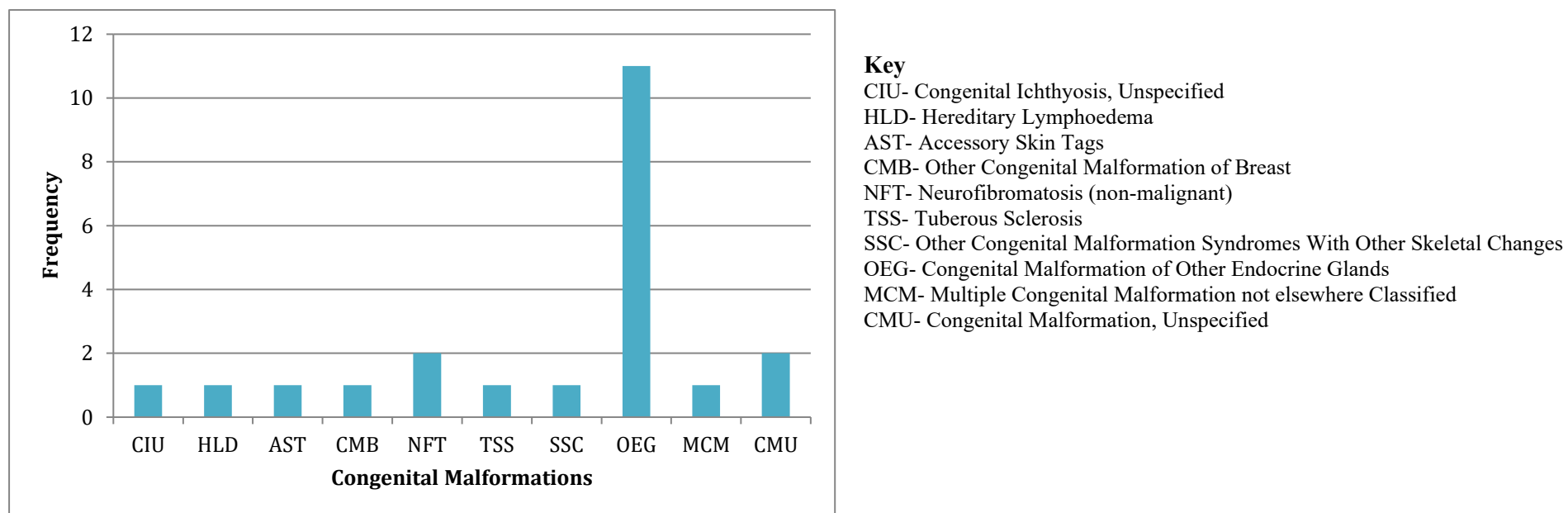


Fig. 9 Bar chart showing the frequency of congenital malformations and deformities of the musculoskeletal system, January 2016–December 2018

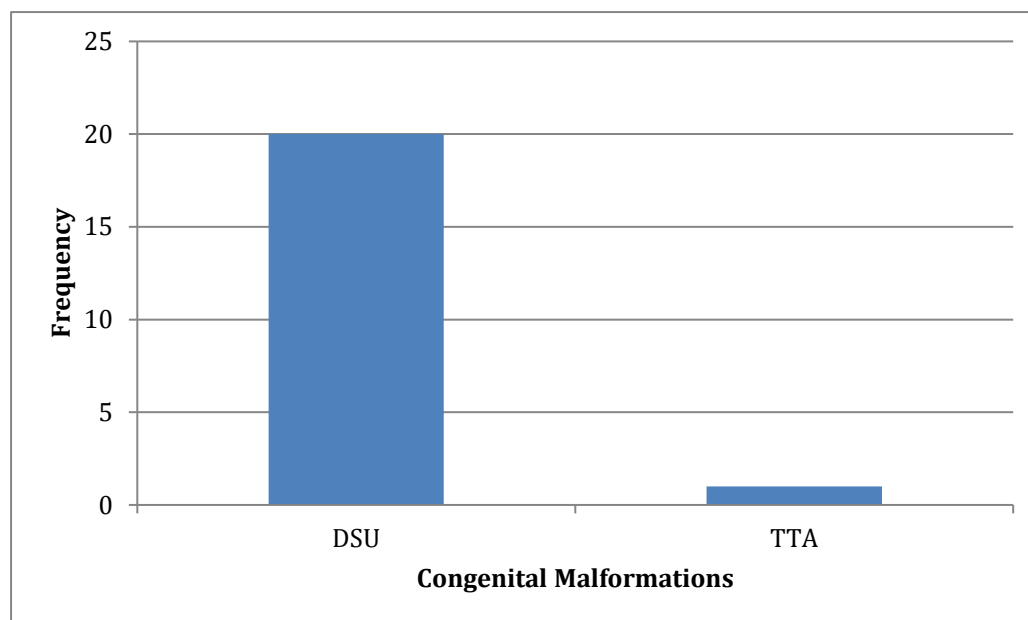


18

19 Fig. 10 Bar chart showing the frequency of other congenital malformations, January 2016–December 2018

20

21



Key

DSU- Down Syndrome, Unspecified

TTA- Trisomy and partial Trisomy of Autosomes, Unspecified

22

23 Fig. 11 Bar chart showing the frequency of chromosomal abnormalities not elsewhere specified, January 2016–December 2018

24

25

Discussion

26 The present retrospective study assessed the frequency of congenital anomalies at the University of Calabar Teaching Hospital
 27 (UCTH), Calabar, from January 2016 to December 2018. A total of 438 cases and 129 anomalies were observed during this period,

28 with talipes equinovarus being the leading anomaly in each year. After data analysis, the observed frequency was found to be
29 statistically significant.

30 The findings are consistent with reports from other Nigerian studies, which have documented varying prevalence rates of congenital
31 anomalies across different regions [7, 8, 21, 22]. The predominance of musculoskeletal anomalies, particularly talipes equinovarus,
32 is in agreement with studies from other sub-Saharan African settings [10, 26]. The high proportion of anomalies recorded among
33 outpatients suggests that many affected neonates were born to mothers who did not receive adequate antenatal care, thereby
34 missing opportunities for early detection through genetic screening and ultrasound scanning.

35 The strong genetic predisposition indicated by the case files for conditions such as congenital heart malformation and congenital
36 cataract is consistent with published literature [27, 24]. Environmental and multifactorial contributions to the aetiology of birth
37 defects, including exposure to air pollution, pesticides, and proximity to hazardous waste sites, have also been documented [5, 6,
38 30] and may be relevant in the context of the study population.

39 **Conclusion**

40 Talipes equinovarus, congenital malformation of the heart, and congenital cataract are the three most prevalent anomalies
41 recorded at UCTH and may be linked to genetic origin. The majority of anomalies were recorded in outpatients rather than
42 inpatients, suggesting that women who do not attend antenatal clinic during pregnancy have a higher chance of delivering a

43 congenitally malformed child, and would not have been informed about preimplantation genetic diagnosis (PGD) testing and
44 ultrasound scans for early detection. Strengthening antenatal care services and establishing a formal congenital anomalies registry
45 at UCTH are recommended to improve surveillance and management of these conditions.

46 **Declarations**

47 **Acknowledgements**

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49 Calabar Teaching hospital, Medical Records Unit, Doctors, and Information Officers for their great assistance in retrieval of the
50 records used in this study. Thank you a great deal.

51 **Author contributions**

52 IOP: Conceptualization, Supervision, Data curation, Formal analysis, Writing – original draft. JOP: Data curation, Writing – review &
53 editing. CDA: Data curation, Investigation.

54 **Funding**

55 The authors jointly funded the research.

56 **Data availability**

57 The datasets used during the current study are not publicly available due to the privacy policies of the Hospital used but are
58 available from the corresponding author on reasonable request.

59 **Ethical approval and accordance**

60 Ethical approval was obtained prior to data collection from the University of Calabar Teaching Hospital Health Research Ethics
61 Committee (UCTH HREC). The approval number was UCTH NHREC REG NUMBER: NHREC07/10/2012 and HREC protocol
62 assigned number: UCTH/HREC/33/684. As this was a retrospective review of existing hospital records, individual patient informed
63 consent was waived by the ethics committee. The research was carried out in accordance with the guidelines and protocols
64 approved by the ethics committee above.

65 **Consent for publication**

66 This study did not include individual patient data that would require consent for publication.

67 **Competing interests**

68 The authors declare no competing interests.

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