

# A Review of Congenital Anomalies Presenting at Arthur Davison Children's Hospital

Isaac Kunda<sup>a</sup>\*, Seter Siziya<sup>b</sup>, Victor Mwanakasale<sup>c</sup>

<sup>*a,b*</sup>Copperbelt University, School of Medicine, Public Health Unit, Ndola, Zambia. <sup>*c*</sup>Copperbelt University, School of Medicine, Basic Sciences Department, Ndola, Zambia.

# Abstract

Disruption of early stages of human development usually happens during the period of organogenesis, which refers to the period between 4<sup>th</sup> and 8<sup>th</sup> week of gestation period. This often leads to developmental anomalies (congenital anomalies), which contribute to mortality and morbidity especially in infants. The aim of this study is to describe the amount (frequency) and pattern of distribution of congenital anomalies presenting at Arthur Davison Children's Hospital. A descriptive retrospective study of 161 cases of congenital anomalies that presented at Arthur Davison children's Hospital was carried out. The data was collected from the hospital's records, and covered the period from January 2015 to December 2015. Congenital anomalies of the central nervous system had the highest occurrence with 65 (40.4%) cases; urogenital system showed 39 (24.2%) cases; musculoskeletal system had 37 (22.9%) cases; gastrointestinal system 13 (8.1%) cases; cardiovascular system showed 2 (1.2%) cases, whereas 5 (3.1%) cases were for multiple anomalies. A significant association between congenital anomalies of the urogenital system and gender was observed (P <0.001). The recorded cases at Arthur Davison Hospital showed various cases of congenital anomalies involving different organ systems. It is therefore, important that careful intrauterine detection and examination of newborns be carried out in order to detect, and possibly correct early cases of congenital anomalies. Antenatal visits still remain an integral part of policies aimed at curbing and reducing the occurrence of congenital anomalies.

Keywords: Congenital anomalies; Arthur Davison Children's Hospital; Tertiary hospital.

-----

\*Corresponding author

## 1. Introduction

A congenital anomaly is any abnormal structural or medical condition that presents at birth [1]. They are often referred to as birth defects. 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]. Major structural anomalies occur in 4-6% of live born babies and are the leading cause of infant mortality and morbidity, accounting for about 25% of deaths, not discriminating on racial grounds [2]. Till the 1940s, it was believed that the human embryo developed in an impervious maternal uterus. It was as such protected from environmental hazards, drugs inclusive, by the amnion and chorion. Hereditary factors were hence thought to be the primary cause of congenital anomalies [1]. 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 [4]. The majority, 40%-60% of congenital anomalies have unexplained causes [5].

The aim of this study is to describe the frequency and pattern of congenital anomalies presenting at Arthur Davison Children's Hospital. The need for quality and better health care service delivery cannot be overemphasized. This research will provide a database for further studies on congenital anomalies, and help in the suggestion of preventive measures in future. This research will also enrich the field of Anatomy with knowledge on the types and pattern of congenital anomalies for the Northern region of Zambia as will be reflected at Arthur Davison children's hospital.

## 2. Materials and Methods Used

## 2.1.1. Study Location

The study was carried out at Arthur Davison Hospital, which is the largest Children's referral hospital in Zambia, located in Ndola, the provincial capital for the Copperbelt province of Zambia. This is a tertiary hospital, and it caters mostly for referrals from the northern region of Zambia which include: Copperbelt, Northwestern, Luapula, Muchinga, Northern, and part of Central provinces of Zambia.

# 2.1.2. Study Type

This is a descriptive retrospective study, which shows the frequency and pattern of distribution of congenital anomalies presenting at Arthur Davison Hospital.

# 2.1.3. Sample Size

A total number of 161 cases of congenital anomalies were recorded for the period from January 2015 to December 2015.

# 2.1.4. Variables

The variables for this research were the: frequency of congenital anomalies by biological systems, pattern of distribution of congenital anomalies, and congenital anomalies of the body systems by gender (sex).

## 2.1.5. Data Analysis

Frequency tables, Pie Chart and Bar chart were used to represent the variables. Calculation for the significant association between congenital anomalies of the urogenital system and gender was carried out. The Fisher Exact Test was used to test for association.

# 3. Results

Out of a total of 161 congenital anomalies recorded, that of the central nervous system was the highest with 40.4%, followed by the urogenital system with an occurrence of 24.2%, whereas unclassified cases represented 3.1% of the study population (Tables 1 and Figure 1).

Anomaly by system	Frequency (n)	Percentage (%)
Cardiovascular system	2	1.2
Central nervous system	65	40.4
Gastrointestinal system	13	8.1
Urogenital system	39	24.2
Musculoskeletal system	37	22.9
Unclassified	5	3.1
Total	161	

# Table 1: All recorded congenital anomalies by body systems



Figure 1: Percentage (%) of congenital anomalies by system





	SEX				
SYSTEM		MALE		FEMALE	
	NUMBER (n)	PERCENTAGE (%)	NUMBER (n)	PERCENTAGE (%)	P-VALUE
CVS	0	0	2	3.8	0.103
CNS	38	34.9	27	51.9	0.058
GIT	8	7.3	5	9.6	0.758
UGS	37	33.9	2	3.8	< 0.001
MSS	24	22	13	25	0.692
UNCLASS	2	1.8	3	5.8	0.323
TOTAL	109		52		

#### **Table 2:** Distribution and frequency of congenital anomalies by sex

## 4. Discussion

A total of 161 congenital anomalies were recorded for the period January 2015 to December 2015. Congenital anomalies of the central nervous system had the highest (40.4%) occurrence. This turn out of cases at the children's referral hospital can be attributed to the fact that surgical management of these anomalies requires highest level of specialization. This, however, is in consistent with the results of a similar research done in Tanzania [6]. Congenital anomalies of the urogenital system were the second highest with an occurrence of 24.2%. 37 cases were recorded in males and 2 cases in females. This is consistent with a similar study done in Atlanta, USA where defects of the sex organs were eight and one-half times more prevalent among males and accounted for about half of the increased risk of birth defects among males relative to females. Urinary tract defects were 62% more prevalent among males [7]. Unclassified cases represented 3.1% of the study population. These cases include cases that were not properly categorized at the point of diagnosis/recording, cases of multiple anomalies that cut across more than one body system that could not be grouped alongside those known. Congenital anomalies of the cardiovascular system are the least occurring, with only 2 (1.2%) cases of ventricular septal defect and ectopic heart being recorded in females respectively. Congenital anomalies of the gastrointestinal system had a frequency of 13 (8.1%). On the other hand, congenital anomalies of the musculoskeletal system are the third most occurring anomalies with the occurrence of 22.9%.

Of all the recorded cases of the types of congenital anomalies, Congenital Hydrocephalus had the highest occurrence of 28.6%, followed by Hypospadias with an occurrence of 17.4%. Cleft palate and Lip scored the third most occurring congenital anomaly for the year 2015 with an occurrence of 13%, while Spina-bifida had a frequency occurrence of 6.8%.

A significant association between congenital anomalies of the urogenital system and gender was observed (P <0.001).

Overall, congenital anomalies of most body systems occurred more in males (67.7%) than in females (32.3%). This can be attributed to an X-linked recessive gene, where males are affected more if they carry a single copy as compared to females who are only affected if they carry two copies of this gene. Because it is more likely that someone will inherit one rather than two copies of an abnormal gene, X-linked diseases and conditions are much more common in males than in females [8].

## 5. Conclusion

In conclusion, there is a fairly high number of cases for congenital anomalies in the northern region of Zambia. This clearly calls for maximized effort on policy formulation and implementation towards curbing the occurrence of congenital anomalies.

#### 6. Limitations of the Study

The major limitation of this study was poor records keeping and data management at Arthur Davison Children's Hospital. This had a negative bearing on this study as the period for data collection had to be extended, to give room for the registry assistants to retrieve records.

## 7. Recommendations

Ultimately, reduction in the mortality and morbidity associated with congenital anomalies is purely an attainable goal [9]. This can be achieved through deliberate policies and primary preventive measures that may see the possible outcome of healthy pregnancies. Examples of programs that can help in curbing the occurrence of congenital anomalies may include: comprehensive periconceptual and pre-pregnancy intake of folic acid fortified food, immunization against debilitating diseases such as rubella, and stringent measures and policies against any risk behavior such as alcohol intake during pregnancy. Therefore, antenatal visits by pregnant mothers should be emphasized, as they are very significant in ensuring a healthy pregnancy, hence reducing on the chances of having some of the congenital malformations. Early intrauterine detection of congenital anomalies by ultrasonic examination and possible preventive and curative measures cannot be overemphasized [10].

Records keeping and data capturing systems ought to be improved at Arthur Davison Children's Hospital. This is cardinal and cannot be overemphasized, as future research depends on organized and accurate hospital records.

## Acknowledgments

We are grateful to the management at Arthur Davison Children's Hospital for according us clearance to review records at the hospital. This gratitude is also extended to the staff in the Registry department at the hospital for their assistance and commitment rendered to us.

# References

- [1]. T.W. Sadler. Congenital malformations. Langman's Medical Embryology. 8th ed. 2000. 95
- [2]. K.L. Moore and T.V.N. Persaud. The Developing Human: Clinically Oriented Embryology. 7th ed, Saunders. Elsevier Science, USA. 2003. 560pp
- [3]. P.L. Williams, L.H. Bannister, M.M. Berry. et al (eds.) Gray's Anatomy, The Anatomical Basis of Medicine and Surgery. 38th ed. Churchill Livingstone, Edinburgh, 1995. 2092pp.
- [4]. K. Nelson and L.B. Holmes. Malformations due to presumed spontaneous mutations in newborn infants. N Eng J Med, 1984, 320: 19-23.
- [5]. R.E. Stevenson. The Genetic Basis of Human Anomalies. In: Stevenson RE, Hall JG, Goodman RM (Eds.), Human Malformations and Related Anomalies. Vol. 1.New York: Oxford University Press. 1993. 115pp.
- [6]. M. Florentina. Pattern and factors associated with congenital anomalies among young infants admitted at Bugando medical centre, Mwanza, Tanzania. 2013. http://www.biomedcentral.com/1756-0500/7/195
- [7]. J.M. Lary and L.J. Paulozzi. Sex differences in the prevalence of human birth defects: a populationbased study, Atlanta, USA. 2001, 237-51.
- [8]. Rick Daniels and Lesli H. Nicoll. Contemporary Medical-Surgical Nursing, 2<sup>nd</sup> edition, Cengage Learning, USA. 2011. 159pp.
- [9]. E.O. Ekwere, R. McNeil, B.P. Agim, B. Jeminiwa, O. Oni, and S. Pam. A retrospective study of congenital anomalies presented at tertiary health facilities in Jos, Nigeria. www.arpapress.com/Volumes/JPCS/Vol3/JPCS\_3\_03
- [10]. Abdul Hamid, Qaiser Inayat, Jahanzaib Khan. Incidence and nature of human congenital anomalies, Peshawar Print, Pakistan. J. Med. Sci. (Peshawar, Print) April 2015, Vol. 23, No. 2: 69-72
- [11]. F.E.A. Lesi. A study of congenital malformations in the newborn in Lagos, Nigeria. Ph.D. thesis University of Dublin,1968.111-124
- [12]. C.J. Chen, C.J. Wang, M.W. Yu. And T.K. Lee. Perinatal mortality and prevalence of major congenital malformations of twins in Taipei city. Acta Genet Med Gemellol (Roma). 1992. 41(2-3): 197-203.