Pattern of congenital anomalies among pediatric surgical patients in a tertiary care hospital in northern Tanzania

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ABSTRACT

Background Congenital anomalies are major causes of morbidity and mortality in children under 5 years of age and make a significant contribution to the surgical burden of diseases. Most anomalies have multifactorial causes and commonly affect the central nervous, cardiovascular, gastrointestinal and musculoskeletal systems. Countries with improved pediatric surgical care have shown dramatic reductions in morbidity and mortality rates. The aim of this study was to analyze the pattern of congenital anomalies presenting in our surgical departments in patients under 5 years of age.

Methods A retrospective descriptive study was done. Data were obtained from clinical records of patients under 5 years of age, who underwent surgical correction of their congenital anomalies between 2017 and 2021. Analysis was done to identify the proportion of congenital anomalies managed in our setting.

Results Congenital anomalies contributed 4.6% of overall surgical burden. Totally, 822 patients with congenital anomalies were included for analysis. The most commonly diagnosed congenital anomaly was inguinal hernia, followed by hydrocephalus, neural tube defects and cleft lips. The most commonly affected system was the central nervous system, anterior abdominal wall, orofacial and digestive system in decreasing order of frequency. Most of our patients presented outside the neonatal period (84.4%), and few (16.1%) had more than one system affected. Male children comprised 64%.

Conclusions Delayed presentation of children with congenital anomalies is still a significant problem in our area. Prevention through nutritional supplementation and antenatal screening is crucial. The true epidemiology of congenital anomalies in northern Tanzania is still obscure.

INTRODUCTION

Congenital anomaly (CA) is a defect in the morphogenesis of an organ that results from defective embryogenesis. CAs affect 1 in every 33 newborns. The global estimated prevalence of CAs is between 1% and 3% and varies widely among countries. Ninety-four percent of CAs occur in low and middle-income countries (LMICs) owing to high fertility rates, low pregnancy termination rates, nutritional deficiencies, increased intrauterine infections, and exposure to teratogens.

For most CAs, the cause is not identified and is considered multifactorial, which accounts for as many as two-thirds of all defects. However, in most African cultures, superstitious beliefs dominate as the cause of these anomalies. Several risk factors have been attributed to the development of CAs. These include metabolic factors, such as hypothyroidism, uncontrolled diabetes, and maternal obesity. In addition, maternal exposures to certain drugs (trimethoprim, phenytoin, phenobarbitone and carbamazepine), infections (TORCHs—t toxoplasmosis, others (syphilis, hepatitis B), rubella, cytomegalovirus (CMV) and herpes simplex) and radiation have been implicated. Similarly,
maternal age (>35 years), family history of CAs, maternal history of CAs, high birth order (>4 births) and consanguineous marriage have been reported as significant risk factors for CAs. Preconceptual multivitamin supplements with folic acid at 400 mcg/day up to the 12th week of gestational age have been shown to reduce the rate of CAs.

CAs contribute a significant proportion of infant mortality globally. It is estimated that they account for one-third of infant mortality, 8%–15% of perinatal deaths and 13%–16% of neonatal deaths. Globally, CAs are the fifth leading cause of death in children under 5 years of age, accounting for about 500,000 deaths annually, 97% of these occurring in LMICs. They account for about 9% of the surgical burden of diseases, and if left untreated, they contribute to the morbidity and mortality of 150 million children globally. They also account for a large number of cognitive and functional disabilities. The morbidity for untreated CAs is estimated at 57.7 million disability-adjusted life-years (DALYs) worldwide.

Up to two-thirds of deaths and disabilities from CAs could be avoided by adequate surgical care. Significant advances in surgical management of CAs have resulted in treatment success in up to 90% of cases. In LMICs, these conditions are often left untreated owing to poor access to surgery, limited community awareness of potential treatment and stigmatization. Surgery is a cost-effective way to mitigate the significant premature mortality and lifelong disability from CAs.

The overall prevalence and pattern of distribution of CAs in Tanzania remains unknown in literature. Several studies in Tanzania focusing on local regions have reported prevalence ranging from 0.28% to 6.05%, with the central nervous system (CNS) being the most common affected system throughout. The paucity of data on CAs is due to poor diagnostic capabilities in most health facilities, lack of awareness on management of CAs and absence of surveillance. This study was conducted with the aim of studying the distribution of various CAs among pediatric patients (under 5 years of age) who underwent corrective surgical treatment between 2017 and 2021.

**METHODS**

A retrospective analysis was conducted in the Departments of Pediatric Surgery, Neurosurgery, and Orthopedics at Arusha Lutheran Medical Centre from July 2017 to June 2021. The study population comprised 822 children aged 5 years and below, who were admitted with CAs. Information on age at presentation and sex was documented. Other significant information like birth order, consanguinity, maternal illness, ingestion of drugs during pregnancy, exposure to radiation during pregnancy, antenatal ultrasonography findings, and mode of delivery was not recorded because most patients lacked this information.

The major malformations were divided into CNS, gastrointestinal tract (GIT)/digestive system, anterior abdominal wall defects (AWD), musculoskeletal, genitourinary (GU), orofacial (OF), and others. Patients with cardiovascular anomalies were excluded because they are not managed in our center but are referred to pediatric cardiac surgeon. We also excluded those patients who were admitted with CAs but did not undergo surgery. Files of the included patients were analyzed to obtain data on the type of anomaly, age of presentation and sex. Data extraction was done using Microsoft Excel, and SPSS V.25 was used for data analysis.

**RESULTS**

CAs in our study contributed 4.6% of the overall surgical burden during the study period. During the study period, 1562 patients under 5 years of age underwent surgery for various reasons. Of these, 822 patients underwent correction for CAs. Most of our patients (64%) were males, and the majority (84.4%) presented outside the neonatal period (see table 1).

Children with more than one diagnosis were 16.1% (n=132), mostly affecting the CNS. The anomalies have been classified according to the system affected (see table 2). The most commonly diagnosed anomaly was inguinal hernia (19%, n=172), followed by hydrocephalus (18.6%, n=169), neural tube defects (NTDs) (11.8%, n=108) and cleft lips (11.5%, n=104). The CNS was the most affected system accounting for 30.4% of all managed defects. These included spinal dysraphisms, encephalocele and hydrocephalus. Hydrocephalus was commonly associated with NTDs (40 patients) and aqueduct of Sylvius stenosis (17 patients) (see table 3). In most patients (n=108), the cause of hydrocephalus was not documented. Spinal dysraphisms were most commonly lumbosacral (n=31), followed by lumbar (n=28), thoracolumbar (n=12) and sacral (n=11). Encephaloceles were commonly occipital (54.5%, n=12) (see table 4).

The next most common defects were the AWDs (26.4%), OF clefts (22.6%) and the digestive system (11.5%). AWDs comprised inguinal hernias (72%), umbilical hernias (23.4%), omphalocele (2.5%), and gastrochisis.

| Table 1 Demographics | Patients (n=822) |
|-----------------------|-----------------|
| **Variables**         | **Patients (n=822)** |
| **Age (mon)**         | **Patients (n=822)** |
| ≤1                    | 128 (15.6) |
| >1 to ≤12             | 430 (52.3) |
| 13–60                 | 264 (32.1) |
| **Sex**               | **Patients (n=822)** |
| Male                  | 526 (64) |
| Female                | 296 (36) |
| **Data are presented as number (percentage).** |
(2.1%). Of inguinal hernias, 48.8% occurred on the right side, 30.2% were bilateral and the remaining on the left side (21%). Of clefts were cleft lips (50.7%), cleft palate (22%), cleft lip and palate (27.3%).

Anomalies of the digestive system included anorectal malformations (ARM, 53%), Hirschsprung’s disease (18.2%), intestinal atresia (12.5%), and others (16.7%) (table 2). ARMs were mostly anterior ectopic anus (n=15), imperforate anus (n=11), rectovaginal fistula (n=11) and rectourethral fistula (n=7) (table 5).

GU anomalies comprised 3.8% of all managed defects. They comprised hypospadias, undescended testicles, posterior urethral valves, and disorders of sexual differentiation. The most common location for hypospadias was penile shaft (66.7%, n=20), followed by coronal (n=5), perineal (n=3) and glandular (n=2).
Other anomalies managed included hemangiomas (n=3), teratoma (n=1), branchial cleft cyst (n=2), cystic hygroma (n=4), and diaphragmatic hernia (n=1).

**DISCUSSION**

Similar to other studies, we found males to account for the majority of the CAs.4 15 16 The reasons for this have not been clearly elucidated yet. In our study, the most common affected system was the CNS.16 However, unlike other studies, we found hydrocephalus as the most common anomaly, whereas others have reported NTDs as the most common anomaly of the CNS.16–18 Different from Adeleye and Olowookere,17 who reported most hydrocephalus being related to aqueductal stenosis, most of our cases of hydrocephalus were related to NTDs. Among NTDs, similar to other studies, we found spinal dysraphisms to be more common than encephaloceles.17 18

Among GIT anomalies, ARM was the most common anomaly accounting for 6.1% of all CAs, which is low compared with the findings of another study done in a different part of the country that reported a rate of 19.6%.15 We failed to obtain a clear reason for this difference. We also found low incidence of associated anomalies.15 This may be attributed to the lack of adequate screening services, such as pediatric echocardiography and reliable ultrasonography, because most of our CAs were diagnosed clinically.

We found isolated cleft lips to be more common than isolated cleft palates and cleft lips and palates combined, similar to another study done in Tanzania by Manyama et al. Several studies in other parts of the world have reported varying prevalence rates of cleft lips and palates.19–21 These differences have been attributed to biological and ethnical differences.19

The majority of our patients (84.4%) presented outside the neonatal period. Delayed presentation to medical facilities is a common occurrence in poor resource countries, Tanzania being no exception. Most of those who presented early had anomalies of the GIT. This late presentation for surgical management of anomalies places a significant burden on DALYs.22 Early presentation and adequate intervention significantly improve the outcome of CAs though it increases healthcare costs.23 24

Prenatal diagnosis of CAs would also significantly reduce the delays in presentation for children with CAs. However, it is the authors’ experience that few women have access to ultrasound screening during the prenatal period due to its unavailability in most primary healthcare facilities. In addition, the focus of the ultrasound is not to assess for CAs. Consequently, those born with CAs come ‘as a surprise’. On the same note, genetic studies to screen and assess CAs are largely unavailable to the public and are available in few research institutions. We found no published studies done in Tanzania that report the availability of prenatal ultrasound screening or genetic studies for CAs.

Although the study location captures most of the cases from northern Tanzania owing to the availability of pediatric surgery and neurosurgical services, the true prevalence of CAs in northern Tanzania is still obscure because many children, for a variety of factors, do not present to health facilities and because this study focused on a single institution.

In conclusion, a significant number of children in our region suffer a wide range of CAs. Management of these anomalies, while associated with increased healthcare costs, provides the child with a chance of normal living. There is a need for the government to increase healthcare funding especially for the pediatric population. Most importantly, simple measures, such as folic acid consumption and/or food fortification in women, can help reduce a lot of this burden (particularly NTDs).

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**Table 3** Hydrocephalus classification according to causes

| Type of cause                           | n (%)   |
|----------------------------------------|---------|
| Unknown                                | 108 (63.9) |
| NTD related only                       | 35 (20.7) |
| Aqueductal stenosis                    | 12 (7.1)   |
| Aqueductal stenosis+NTD                | 5 (3)    |
| Dandy-Walker malformation              | 4 (2.3)    |
| Aqueduct blockage                      | 5 (3)    |
| NTD, neural tube defect.               |          |

**Table 4** Neural tube defect classification

| Type of neural tube defect                   | n (%)   |
|---------------------------------------------|---------|
| Lumbosacral                                 | 31 (28.7) |
| Lumbar                                      | 28 (25.9) |
| Thoracolumbar                               | 12 (11.1) |
| Sacral                                      | 11 (10.2) |
| Thoracic                                    | 4 (3.7)   |
| Occipital encephalocele                     | 12 (11.1) |
| Frontal encephalocele                       | 5 (4.6)   |
| Frontonasal encephalocele                   | 5 (4.6)   |

**Table 5** Types of anorectal malformations

| Type of malformation                        | n (%)   |
|---------------------------------------------|---------|
| Anterior (ectopic) anus                     | 15 (27.3) |
| Imperforate anus                            | 11 (20)  |
| Rectovaginal fistula                        | 11 (20)  |
| Rectourethral fistula                       | 7 (12.7) |
| Cloaca                                       | 4 (7.3)  |
| Perineal fistula                            | 2 (3.6)  |
| Anal stenosis                               | 2 (3.6)  |
| Unclassified                                | 3 (5.4)  |
Prevention is the most important form of treatment for CAs.

Limitations
This study has several limitations, namely its retrospective nature which hinders access to pertinent information. Exclusion of data pertaining to factors associated with CAs also limits the potential of the study to assess potential risk factors in our set-up. In addition to the above, exclusion of patients above 5 years of age means other patients with CAs were not assessed.

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All data relevant to the study are included in the article or uploaded as supplementary information.

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