Profile of Major Congenital Anomalies Seen at University Teaching Hospital, Lusaka: A Petition for Congenital Anomalies Surveillance

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ABSTRACT

Objectives: To describe the profile of congenital anomalies at University Teaching Hospital (UTH), Lusaka with special reference to implications for training and integrating congenital anomalies prevention strategies in primary health care.

Design: The profile of congenital anomalies was determined by reviewing research records that had been collected over a period of 20 years (January 1985 – January 2005).

Results: The total incidence of major congenital anomalies was 1.6% (n = 5,478) of the patients who attended the neonatal and paediatric surgical wards during the period under study. The categories of defects with the highest prevalence were digestive system anomalies (38.9%), nervous system anomalies (31.5%), cleft lip and palate (12.3%) and urogenital anomalies (9.7%). Of note was the relative low contribution of congenital heart disease (1.7%). The commonest digestive system anomalies were severe aganglionosis (n = 701), ano-recto atresia (n = 501), jejenual and duodenal atresias (n = 270) and exomphalos (n = 101); The central nervous system anomalies included spina bifida (n = 1,500), encephalocoele (n = 121); those for orofacial clefts were cleft lip and palate (n = 670); and for the urogenital system congenital hydronephrosis (n = 210), ectopic bladder (n = 99) and ambiguous genitalia (n = 75). Other congenital anomalies recorded included osteogenesis imperfecta, achondroplasia, congenital dislocation of the hip, patent ductus arteriosus, ventricular septal defect, tricuspid atresia, Down's syndrome and Pierre Robin syndrome to list a few.

Conclusions: Major congenital anomalies are prevalent in Zambia, and the incidence may be increasing. The considerable challenge posed by major congenital malformations in Zambia arouses the need for the establishment of hospital Congenital Abnormalities Registry and Surveillance System (CARSS) and its inclusion on the Health Monitoring Information System(HMIS), and ultimately the development of prevention programmes through the integration of preventive measures into primary health care and maternal and child health services.

INTRODUCTION

In Zambia the control of genetic disorders and congenital anomalies is generally not given the importance it deserves. Many, wrongly, believe that the problem is minor in magnitude and that little can be done about it. This article aims to provide a brief outline of the magnitude and types of major congenital abnormalities seen at University Teaching Hospital (UTH), the national referral centre for Zambia. The information can benefit national health planners, researchers and medical educators. Additionally, the article petitions for the establishment of hospital and a national Congenital Abnormalities Registry and Surveillance System (CARSS) and for the inclusion of congenital anomalies on the Health Monitoring Information System (HMIS) that is used monitor diseases in Zambia.

Epidemiological Situation

Little is known and/ or documented about the epidemiological situation of congenital anomalies in Zambia. However, worldwide, genetic disorders and congenital anomalies occur in 2%-5% of all live births and account for almost 30% of paediatric hospital admissions. They cause about 50% of childhood deaths in industrialised countries[1]. Sherry[2] asserts that major birth defects or congenital anomalies occur in approximately 8.3 percent of all live births. The reported

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incidence of congenital anomalies vary, for example, in the United States spina bifida is reported at 1 in 1000 live births, while it is 1 in 250 to 500 live births in Great Britain and South Africa[2]. Canada reports the following annual prevalence rates: Down's syndrome 1 in 800 live births; neural tube defects (NTDs) 5.8 per 10,000 total births; congenital heart defects (CHDs) 1 in 104 per 10,000 newborns[3]. Countries in Europe also have different rates for most incidences but many are comparable[4].

Preliminary findings, from working experience, are suggestive of an increase in congenital malformations seen at UTH which could be attributed lack of public health measures directed at the prevention of congenital and genetically determined disorder, and inadequate health care prior to and during pregnancy. Perhaps, HIV and AIDS have a hand in this too.

The availability of basic epidemiological data on congenital anomalies is critical to national health planning. The data and capacity thereof empowers the system with the ability to detect changes, investigate the changes, and plan appropriate ad hoc interventions as well as developing appropriate health service policies.

MATERIALS AND METHODS

Patients attending University Teaching Hospital (UTH) neonatology and paediatric surgical wards between January 1985 and January 2005, with a diagnosis of any major congenital anomaly, were prospectively recorded into research records for the department. A retrospective analysis of those records was done for this article. Only major congenital anomalies were included for this study. Where multiple anomalies existed in the same patient the anomalies were recorded under each system, for example, tracheo-oesophageal atresia would be recorded in both respiratory and gastrointestinal systems. Still births were excluded from this record. Where required, cases were evaluated by plain and special x-rays, ultrasound, CT-scan and angiography, before entry into the research records. Only a few post-mortems were done due to difficulty with consent from custodians.

RESULTS

One million five hundred and twelve thousand patients attended the neonatal and paediatric surgical wards during the period under study. Of these 5,478 were diagnosed with a major congenital anomaly representing a 1.6 percent incidence of attendants to neonatal and paediatric surgical wards.

Table 1 shows the distribution of major anomalies attending UTH neonatal and surgical paediatric wards from January 1995 – January 2005 according to the group/system(s) affected.

Sub-group Classification/System(s) Affected	Total Frequency	Percentage of Total Major Malformations (5,478)
Digestive system anomalies	2131	38.9
Nervous system anomalies	1723	31.5
Cleft lip and palate	670	12.3
Urogenital anomalies	536	9.7
Musculoskeletal defects	173	3.2
Cardiac defects	95	1.7
Respiratory system defects	69	1.2
Others	81	1.5
Total	5,478	100

Table 1

The ages of the patients ranged from a few hours after birth to one month, and averaged seven days. Fifty five percent of the patients were female. The commonest malformations are shown in table 2 below.

Sub-group Classification/System(s) Affected	Pathology (Diagnosis)	Frequency
Digestive system anomalies	Severe aganglionosis	701
	Ano-recto atresia	501
	Jejunal & duodenal atresia	270
	Exomphalos	101
Nervous system anomalies	Spina bifida	1500
	Encephalocoele	121
Orofacial Clefts	Cleft lip and palate	670
Urogenital anomalies	Congenital hydronephrosis	210
	Ectopic bladder	99
	Ambiguous genitalia	75
Musculoskeletal defects	Osteogenesis imperfecta	59
	Achondroplasia	36
	Dislocation of the hip	29
Cardiac defects	Patent ductus arteriosus	39
	Ventricular septal defect	15
	Tricuspid atresia	13
Respiratory system	Cystic hygroma (neck)	43
	Pulmonary cyst	11
	Tracheo-oesophageal fistula	19
Others	Down's syndrome	25
	Pierre Robin syndrome	19
	Adrenogenital syndrome	18

Table 2

DISCUSSION

The results of this study demonstrate that major congenital anomalies are prevalent in Zambia and a myriad of types of anomalies were seen at UTH (Figure 1, 2).

Figure 1, Omphalocoele

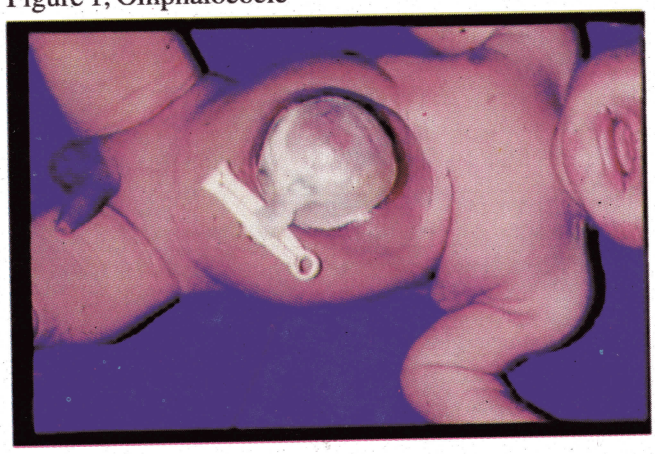
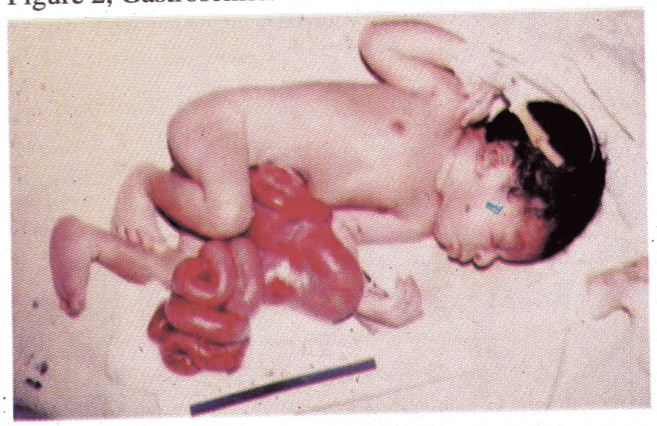


Figure 2, Gastroschisis



The occurrence could even be higher given that no stillbirths and prenatal diagnoses were included in this study. The implications of these findings are that first, our training programmes for health professionals should have enough focus on congenital anomalies to ensure competence in handling them. Second, we need to improve screening of congenital anomalies for early detection and interventions. Thirdly, we need to create systems primarily responsible for collecting incidence data about birth defects. Lastly we need public health measures specifically directed at prevention of congenital and genetically determined disorders to be established.

Collecting accurate data about the geographical and temporal distribution of birth defects is the first step in determining causes and beginning prevention. Invariably, the use of the data is limited to estimates of incidence of birth defects; detecting geographical and temporal shifts in these incidences; detecting 'epidemics' of specific anomalies that might be due to certain environmental agents; and providing useful data for finding the causes of and/or preventatives of birth defects. Subsequently, preventive strategies can be integrated into primary health care, as part of maternal and child health services.

All the above concerns and strategies can be addressed by the creation of Congenital Abnormalities Registry and Surveillance System (CARSS) and its inclusion on the Health Monitoring Information System. The objectives of CARSS could include:

- To provide essential epidemiological information on congenital anomalies in Zambia.
- To facilitate the early warning of teratogenic exposures.
- To evaluate the effectiveness of primary prevention.
- To advocate for the improvement of prenatal screening.
- To aet as an information and resource centre regarding clusters or exposures or risk factors of concern.
- To provide readily collaborative network and infrastructure for research related

to the causes and prevention of congenital anomalies and the treatment and care of affected children.

- To act as a catalyst for the setting up of registers throughout Zambia collecting comparable and standardized data.

Such surveillance, in other parts of the world have resulted in the reduction of congenital anomalies related to advanced parental age, for example Down's syndrome and autosomal dominant conditions; reduction in the occurrence of congenital abnormalities such a neural tube defects by offering folic acid, and avoiding the sequelae of micronutrient deficiencies by promoting healthy nutrition for women, prevention of congenital rubella syndrome by immunizing against rubella; reduction of congenital abnormalities and stillbirths by better control of maternal diabetes, reduction of congenital abnormality and fetal retardation through campaigns for the avoidance of smoking and alcohol intake during pregnancy, and detection and prompt treatment of certain infections such as syphilis.

CONCLUSION

Major congenital anomalies are prevalent in Zambia, and the incidence may be increasing. The considerable challenge posed by major congenital malformations in Zambia calls for establishment of a hospital and a national Congenital Abnormalities Registry and Surveillance System (CARSS) and the inclusion of congenital anomalies on the Health Monitoring Information System that monitors disease in the country. Additionally, the development of prevention programmes and ultimately the integration of preventive measures into primary health care and maternal and child health services are called for. The strategies proposed in this article do not necessarily require complex logistical and technical capabilities but are primarily based on strengthening training of health professionals, public education and the Health Monitoring and Information System with regard congenital anomalies.

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