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Congenital anomalies in Ibadan, Nigeria

AI Ayede[1], AO Adeleye[2], AA Olusanya[3], TA Lawal[4], BA Olusanya[5], OO Ogunkunle[1], AO Asinobi[1], BE Adebayo[1], OO Jarrett[1], DI Olulana[4], OO Ogundoyin[4], OF Ashubu[1], IO Morhason-Bello[6], AOA Adekanbi[6], AM Baiyeroju[5], VI Joel-Medewase[7], AD Ademola[1], O Olayemi[6], GI Ogbole[8], JA Akinmoladun[8], AM Agunloye[8], OO Akinrinoye[1], AO Takure[9], OB Oyewole[1], OM Oluwatosin[10] and SI Omokhodion[1]

Departments of Paediatrics[1], Neurological Surgery Unit[2], Oral and Maxillofacial Surgery[3], Paediatric Surgery Unit[4], Ophthalmology[5], Obstetrics and Gynaecology[6], College of Medicine, University of Ibadan, Department of Paediatrics and Child Health[7], LAUTECH Teaching Hospital, Ogbomoso, Departments of Radiology[8], Surgery, Urology Unit[9] and Plastic Surgery Unit[10], College of Medicine, University of Ibadan, Nigeria.

Abstract

Background: There is a need for a multidisciplinary database that can be used as a potential source for developing a protocol and a guideline for a possible nationwide prospective surveillance of congenital anomalies in Nigeria.

Methods: This five-year cross-sectional retrospective survey of data from January 2009 to December 2013 was done at the University College Hospital, Ibadan. Data were collected from the admission records on the wards in 8 specialty units with the most workable documented clinical records of congenital anomalies in their care using a predesigned proforma. Proportions of congenital anomalies were determined based on systemic classification of the anomalies and the descriptive terms used were according to the ICD_10-chapter XVIII_RCPCH extension.

Result: The total number of patients with congenital anomalies whose records were obtained from the ward registers was 1311, there were 75 (5.7%) missing case notes while information was obtained on 1236 (94.3%) patients. There were a total of 1479 anomalies with multiple anomalies seen in 16.1% of the patients. The male/female ratio was 2:1 and multiple births as well as positive family history of birth defects were seen in about 2.4% and 2.2% of cases respectively. Prenatal diagnosis of the anomalies was documented in only 11 cases (0.9%). Only about one in five cases presented within the neonatal period, and defects of the abdominal wall as well as the cranial-facial-orbital regions were the most prevalent. Next were those in the cardiovascular, spinal column, anorectal and genital, as well as musculoskeletal systems.

Conclusion: The size and pan-systemic profiles of the birth defects documented in this study calls for further action on this all-important cause of childhood mortality and significant life-long morbidity in our country.

Keywords: *Congenital, anomaly, Ibadan*

Résumé

Contexte: Il est nécessaire de disposer d'une base de données multidisciplinaire pouvant être utilisée comme source potentielle pour l'élaboration d'un protocole et d'une ligne directrice pour une éventuelle surveillance nationale prospective des anomalies congénitales au Nigeria.

Méthodes: Cet examende cinq anstransversal rétrospectif sur les données de janvier 2009 à décembre 2013 a été effectué auCollège HospitalierUniversitaire d'Ibadan. Les données ont été recueillies à partir des dossiers d'admission dans les salles dans 8 unités de spécialité avec les dossiers cliniques documentés les plus réalisables des anomalies congénitales dans leurs soins en utilisant un proforma pré-conçu. Les proportions d'anomalies congénitales ont été déterminées sur la base de la classification systémique des anomalies et les termes descriptifs utilisés étaient selon l'extension ICD_10-chapitre XVIII_RCPCH.

Résultat: Le nombre total de patients présentant avec des anomalies congénitales dont les dossiers ont été obtenus à partir des registres de salle était de 1311, il y avait 75 (5,7%) cas manquants, alors que des informations ont été obtenues sur 1236 (94,3%) patients. Il ya eu un total de 1479 anomalies avec des anomalies multiples observées chez 16,1% des patients. Le rapport hommes / femmes était de 2: 1 et les naissances multiples ainsi que les antécédents familiaux positifs de défauts de naissance ont été observés dans environ 2,4% et 2,2% respectivement

Correspondence: Dr. A.I. Ayede, Department of Paediatrics, College of Medicine, University of Ibadan, Nigeria. E-mail: idayede@yahoo.co.uk

des cas. Le diagnostic prénatal des anomalies a été documenté dans seulement 11 cas (0,9%). Seulement environ un cas sur cinq présentait dans la période néonatale, et les défauts de la paroi abdominale ainsi que les régions crânio-faciales-orbitales ont été les plus répandues. Ensuite étaient ceux dans le cardiovasculaire, colonne vertébrale, anorectal et génital, aussi bien que les systèmes musculo-squelettiques.

Conclusion: La taille et les profils pan-systémiques des malformations congénitales documentées dans cette étude appellent une action supplémentaire sur cette cause tout aussi importante de mortalité infantile et de morbidité significative à long terme dans notre pays.

Mots-clés: *Congénitale, anomalie, Ibadan*

Introduction

Birth defects (synonym, congenital anomalies) are structural or functional abnormalities present at birth, and may be inherited or be the result of environmental influence [1,2]. Although the aetiology of most of these defects is unknown, it is estimated that 1 in 33 infants are born with congenital anomalies yearly [3]. More specifically, the incidence of congenital anomalies worldwide is estimated to be about 2.5% - 3.5% in the United States of America, 2-2.5% in the Middle East, 2.5% in India and 1.3% in China [3]. In all, about 270,000 babies worldwide die every year during the first 28 days of life, or about 3 million under the age of five, from these defects [1,4,5]. Furthermore, congenital anomalies may have long term effects on the quality of life of affected individuals, their respective families, and the society in general such as psychosocial affectations and cost implications of management [1,6,7].

Registration of Birth Defects (BDs) is well documented in the developed countries where their surveillance is ensured by highly functional and dynamic epidemiological systems such as the European Network for the Surveillance of Congenital Anomalies (EUROCAT) [8]. On the other hand comparable data from the majority of the African continent, especially the sub-Saharan region, is scarce. Some reports from Africa have documented the prevalence of congenital anomalies as 76.1 per 1000 [5]. Like in most parts of sub-Saharan Africa (SSA) there is currently no BDs surveillance system in Nigeria. Most available reports on congenital anomalies are retrospective, system-specific, hospital-based studies by respective sub-specialty teams. Hence the real burden of congenital anomalies, their trend(s) and the particular population(s) at risk (if any) remain only crudely characterised at best [9-14].

The exact reasons for the scarcity of data from developing countries can be putative but some of the more easily imagined implicating factors include absence of BD registries, lack of positive health seeking behaviours and weak health systems in general [3,4]. Therefore, it is unclear if prevalence values of congenital anomalies from developed countries are similar to those of the developing countries or otherwise. In addition to the factors above, poor nutrition, high prevalence of infectious diseases, high availability of a wide range of over-the-counter prescription-only drugs as well as native concoctions may be responsible for possible differences in the prevalence of these anomalies [4].

Reliable data from registration and surveillance of congenital anomalies may potentially aid research on their actiology and diagnosis with subsequent reduction in disease burden. It would also help in establishing baseline data, documenting changes in rates and fostering international collaboration on the subject. In addition, such data will create an enabling platform needed in implementation of health prevention strategies and policy making towards improving health care status of children in large developing countries like Nigeria [1,5,15]. These benefits are even more important for developing countries as majority of infants with serious congenital anomalies are born in the low or middle income countries where comprehensive records are not available [3,5].

This study therefore aims to provide an initial exploratory survey on the burden of birth defects, in a format comparable to global reports, from the premier tertiary university teaching hospital in Nigeria. It is hoped that this work together with other sub-specialty detailed BD publications to follow would provide a multidisciplinary database that can be used as a potential source for developing a protocol and a guideline for a possible nationwide prospective surveillance of congenital anomalies in Nigeria.

Materials and methods

Study location

This study was done at the University College Hospital, Ibadan, Nigeria; an 850-bed university teaching hospital with more than 60 clinical departments. The hospital is located in the metropolis of Ibadan, a densely populated city in Nigeria, West Africa. The hospital serves as a major referral centre for not only all the population of its regional South West Nigeria, but indeed the whole country at large.

Study design
It is a five-year cross-sectional retrospective survey of data from January 2009 to December 2013.

Study participants
All patients presenting with BDs, born either at the hospital or elsewhere but referred to the University College Hospital, Ibadan and had their records available were recruited into the study.

Acquisition of data
Data were collected from the admission records on the wards in 8 units with the most workable documented clinical records of congenital anomalies in their care: Neonatal, Paediatric Surgery, Paediatric Endocrinology/Genetics, Paediatric Nephrology, Neurosurgery, Cleft clinic, Ophthalmology and Radiology departments. A congenital anomaly was defined as any structural abnormality present at birth; diagnoses of syndromes were made based on clinical examination. Results of chromosomal analyses, echocardiography and Computed Tomographic Scans (CTS)/ Magnetic Resonance Imaging (MRI) were available for few cases in addition to the physical examinations and basic clinical data.

Data management
Case file numbers of the infants were extracted from ward registers of the above units and departments and inputted into a computer using IBM SPSS® version 21. Duplications of records due to possible review of patients by more than one unit were prevented by computing batches of extracted names and hospital numbers on SPSS and checking for recurrence. Detected duplicated cases had one of such cases deleted before retrieval of the concerned case notes. The respective case notes were retrieved from the medical records of each unit and department. Data was retrieved from case notes using a predesigned proforma. Double data entry and consistency checks were done using Epi Data version 3.1. Transfer of clean data to data version 12 was done for final data analysis. Proportions of congenital anomalies were determined based on systemic classification of the anomalies and the descriptive terms used were according to the ICD_10-chapter XVIII_RCPCH extension [16].

Results
The total number of patients with congenital anomalies whose records were obtained from the ward registers was 1311, there were 75 (5.7%) missing case notes while 1236 (94.3%) children had their case notes retrieved and all analyses were based only on these patients. There were a total of 1479 anomalies with 16.1% (n = 99) of the patients having more than one anomaly. The proportion with multiple anomalies, demographic characteristics and type of anomalies are as shown on tables 1, 2 and 3 respectively.

Table 1: Categories of defects

Number of anomaly	Frequency (%)
1	1037(83.9)
2	138(11.2)
3 and more	61(4.9)

Table 2: Demographic characteristics of the patients with congenital anomalies

Variable	Frequency N=1236	Percentage (%)
Age at presentation		
1-23hrs	106	8.6
1-28days	208	16.8
29days-1year	368	29.8
>1 year	554	44.8
Sex		
Male	771	62.4
Female	392	31.7
Ambiguous	7	0.6
Not Reported	66	5.3
Family History of birth defects		
Yes	27	2.2
No	952	77.0
Not Reported	257	20.8
When diagnosed		
Prenatal	11	0.9
Postnatal	1027	83.1
Not Reported	198	16.0
Surgery		
Yes	763	61.7
No	384	31.1
Not Reported	89	7.2
Outcome		
Discharged	1078	87.2
Dead	61	4.9
DAMA*	18	1.5
Not Reported	79	6.4
Parents related		
Yes	2	0.2
No	821	66.4
Not Reported	413	33.4
Multiple birth		
Yes	29	2.4
No	926	74.9
Not Reported	281	22.7

DAMA*: Discharged Against Medical Advice

Table 3: Showing the proportion of patients with the three most common anomalies per system.

Anomalies	Frequency N=1236	Percentage (%)
Respiratory System		
Trachea oesophageal fistula	7	0.56
Congenital cysts	3	0.24
Pulmonary hypoplasia		
Cardiovascular System		
Ventricular septal defect	50	4.04
Atrio-ventricular septal defect	45	3.64
Tetralogy of Fallot	18	1.45
Gastrointestinal System		
Orofacial cleft	119	9.63
Anorectal malformation	51	4.13
Exomphalos	27	2.18
Central Nervous System		
Craniospinaldysraphism	63	5.10
Congenital hydrocephalus	60	4.85
Microcephaly	14	1.23
Endocrine/Genetics		
Down Syndrome	38	3.07
Marfan Syndrome	12	0.97
Ambiguous genitalia	7	0.57
Anterior Abdominal Wall Defect		
Hernias	189	15.29
Hydrocele	100	8.09
Omphalocele	27	2.18
Genitourinary System		
Undescended testes	62	5.02
Posterior urethral valve	51	4.13
Hypospadias	27	2.18
Congenital Eye Defects		
Congenital cataracts	109	8.82
Congenital glaucoma	40	3.24
Strabismus	22	1.78
Musculoskeletal System		
Talipes	9	0.73
Polydactyly	9	0.73
Sacrococcygealteratoma	6	0.49
Other Classifications		
Congenital infection (congenital rubella syndrome)	10	0.81
Ear, nose and throat	6	0.49
Integumentary	3	0.24
Vascular (Haemangioma)	3	0.24
Total	1187*	96.36*

Note* 49 patients (3.64%) had other rare conditions details of which will be presented in the sub specialty manuscripts.

Discussion

In this report, the findings of a 5-year retrospective cross sectional survey of the in-hospital admission cases of congenital anomalies is presented. The study was a hospital-wide multidisciplinary one conducted at a premier university teaching hospital in a large sub-Saharan African country. About 1,479 birth defects in 1236 patients were recorded, male/female ratio of 2:1. Multiple anomalies occurred in (199) 16.1%, multiple births and positive family history of BD were seen in about 2.4% and 2.2 % of cases respectively. Prenatal diagnosis of the anomalies was documented in only 11 cases (0.9%). Only about one in five cases presented within the neonatal period, and defects of the abdominal wall as well as the cranial-facial-orbital regions were the most prevalent. Next were those in the cardiovascular, spinal column, anorectal and genital, and musculoskeletal systems. These proportional distributions of BD in the body systems of our study population agree with similar regional reports in the literature [17,18]. It is also similar in major respects with an autopsy-based study of the same subject earlier carried out in our institution [19].

Two particular points are noteworthy by the pan-systemic nature of the prevalence of BDs in this study. Firstly, BDs are indeed a significant public health problem in the developing countries [20-25], considering that hospital based surveys like this, at best, reveal only a tip of the iceberg. As a matter of fact, the particular 5-year time frame covered by this survey was one in which there were frequent, long-lasting industrial strike actions yearly by various public hospital staff unions during a socio-political crisis period of our country. Hence, what is seen here of the hospital presentation of BD in our university hospital must be only a fraction of what actually exists. Secondly, there is need for national, even regional, multispecialty tertiary health facilities in the developing countries for the comprehensive, multidisciplinary, highly skilled health care needs of patients with congenital anomalies. The latter not only for the immediate clinical care of index cases but more importantly for the development of empiric epidemiological database towards aetiological and diagnostic surveillance, and prevention of these devastating defects. More than 60% of the study subjects were documented to have had surgical treatment of their anomalies, and almost 90% were successfully discharged from the hospital. This is perhaps another poignant attestation of the level of health care possible even in the low-resourced developing countries when there is optimal complement of high level multispecialty personnel present. It also supports the suggestion that most BDs presenting in this part of the world do indeed lend themselves easily to surgical correction and or mitigation [26].

One further interesting finding of this study is the low rate of multiple gestation, 2.4%, among the respective index cases. We could not explain this finding from this study. It would be expected that multiple gestation might predispose to high birth prevalence of congenital anomalies, especially the orthopaedic ones. Perhaps some of the cases did not present in the formal health care setting. Orthopaedic cases like talipes and polydactyly may be considered minor and not serious enough by the parents to warrant hospital visit.

Another noteworthy finding of this study is the low rate of consanguinity in relation to the presence of BDs in the study subjects. However, this fact is in keeping with the cultural backdrop of our study population: marriage among close relations, including cousins (first, second or third, or of any order whatever) is a near inviolable taboo.

Yet another observation from this study is the very low level of prenatal diagnosis and also delayed hospital presentation of BDs. These findings reflect the general trends in the little literature available on the subject in low resource practice areas like our own. They are a further illustration of the suboptimal state of the health systems in these regions [17].

This study suffers from some significant limitations. Most of these were imposed by its retrospective nature. Hence critical parts of the database which might have helped in further characterising the clinical epidemiology of the disease were missing in the not-well documented hospital records. This problem, of highly defective hospital records, is a continuing challenge to academic research in most developing countries [22]. The study is also hospital-based, but it was carried out in the nation's premier university teaching hospital where the most complex/advanced disease conditions are referred. Selection bias to the cases of BDs documented in this study cannot be ruled out, making it unrealistic to make population-wide assumptions on the basis of the findings.

In conclusion, the size and pan-systemic profiles of the BDs documented in this study calls for further action on this all-important cause of childhood mortality and significant life-long morbidity in our country. The results from this study will be found useful in planning for implementation of BD surveillance registry which is currently advocated for in Nigeria and other developing world particularly the Sub-Saharan Africa [27,28].

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