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Congenital paediatric surgical cases in Ibadan: patterns and associated malformations

TA Lawal[1,2], AO Adeleye[1,3], AI Ayede[4,5], OO Ogundoyin[1,2], DI Olulana[1,2], AA Olusanya[6,7], BA Olusanya[8,9], OO Jarrett[4,5], AO Asinobi[4,5], OF Ashubu[5] and OO Busari[5]

Department of Surgery[1], College of Medicine, University of Ibadan, Division of Paediatrics Surgery[2], University College Hospital, Ibadan, Department of Neurological Surgery[3], University College Hospital, Ibadan, Department of Paediatrics[4], College of Medicine, University of Ibadan Department of Paediatrics[5], University College Hospital, Ibadan, Department of Oral and Maxillofacial Surgery[6], College of Medicine, University of Ibadan, Department of Oral and Maxillofacial Surgery[7], University College Hospital, Ibadan, Department of Ophthalmology[8], College of Medicine, University of Ibadan, Department of Ophthalmology[9], University College Hospital, Ibadan, Nigeria

Abstract

Background: There is paucity of data in the developing countries on the outcome of care of children with congenital anomalies managed in a general paediatric surgical setting. The aim of the study was to describe the pattern of congenital anomalies seen in a single tertiary hospital in Nigeria, highlight associated malformations and evaluate in-hospital outcome of care.

Methods: This was a retrospective review of patients who had congenital anomalies of general paediatric surgical nature and had surgery at the hospital over a five-year period. Information was retrieved on sociodemographic characteristics, types of anomalies, associated defects, and in-hospital outcome.

Results: Some 540 out of 1,539 patients, 419 (77.6%) males, operated during the period had congenital anomalies. Their ages ranged from 1 day to 23 years (median, 17 months); the majority (58.7%) presented after the first year of life. The most prevalent anomalies were anorectal malformations (51), exomphalos (27) and hypospadias (27). Patients with respiratory anomalies and anterior abdominal wall defects presented earlier compared to others ($p < 0.001$). Associated major lesions were mostly seen in patients with respiratory (63.6%), anterior abdominal wall (27.6%) and lower gastrointestinal (24.2%) anomalies. Nineteen patients died overall; in-hospital mortality was poorest in those with tracheoesophageal fistula.

Conclusion: Birth defects accounted for 35% of the operative workload of this general paediatric surgery unit. The major congenital anomalies seen were in the gastrointestinal and genitourinary systems; most patients presented late and outcome was worst among patients with tracheoesophageal fistula.

Correspondence: Dr. A.O. Adeleye, Department of Surgery, College of Medicine, University of Ibadan, Nigeria. E-mail: femdoy@yahoo.com

Keywords: *Associated malformations; birth defects; congenital anomalies; general pediatric surgery; in-hospital outcome; developing country*

Résumé

Contexte: Dans les pays en voie de développement, il existe peu de données sur les résultats des soins prodigués aux enfants présentant avec des anomalies congénitales dans un cadre général de chirurgie pédiatrique. Le but de l'étude était de décrire le profil des anomalies congénitales observées dans un seul hôpital tertiaire au Nigeria, de mettre en évidence les malformations associées et d'évaluer les résultats hospitaliers des soins.

Méthodes: Il s'agissait d'un examen rétrospectif de patients présentant avec des anomalies congénitales de nature chirurgicale pédiatrique générale et opérés à l'hôpital, sur une période de cinq ans. L'information a été recueillie sur les caractéristiques sociodémographiques, les types d'anomalies, les défauts associés et les résultats hospitaliers.

Résultats: Quelque 540 sur 1 539 patients, 419 (77,6%) mâles, opérés au cours de la période avaient des anomalies congénitales. Leurs âges variaient de 1 jour à 23 ans (médiane, 17 mois); La majorité (58,7%) a présenté après la première année de vie. Les anomalies les plus fréquentes étaient les malformations ano-rectales (51), les exomphales (27) et les hypospadias (27). Les patients présentant avec des anomalies respiratoires et des anomalies de la paroi abdominale antérieure présentaient plus tôt que les autres ($p < 0,001$). Les lésions majeures associées ont été surtout observées chez les patients souffrant d'anomaliesrespiratoires (63,6%), de murs abdominaux antérieurs (27,6%) et de gastro-intestinalesinférieures (24,2%). Dix-neuf patients sont morts dans l'ensemble; La mortalité intra-

hospitalière était la plus faible chez les patients ayant une fistule de trachée-œsophagienne.

Conclusion: Les anomalies congénitales représentaient 35% de la charge de travail de cette unité générale de chirurgie pédiatrique. Les principales anomalies congénitales observées étaient dans les systèmes gastro-intestinaux et génito-urinaires; La plupart des patients ont présenté tardivement et le résultat était le plus mauvais parmi les patients avec la fistule de trachée-œsophagienne.

Mots-clés: *Malformations associées; malformations congénitales; anomalies congénitales; Chirurgie pédiatrique générale; Résultats hospitaliers; Pays en voie dedéveloppement*

Introduction

Congenital anomalies represent a group of defects that are seen at birth or sometimes later in life. They can be major or minor depending on the severity of the functional and or cosmetic deficits in the organ-system involved. They affect the developing foetus usually at an early stage of embryogenesis, hence there could be involvement of more than one organ-system [1].

In some developing countries, birth defects account for about 2% of overall admissions to children's wards [2, 3] and 24% to 96% of paediatric surgical admissions or operations [4-7]. The most commonly treated major congenital anomalies in general paediatric surgical practice include gastrointestinal tract lesions such as anorectal malformations and Hirschsprung disease, anterior abdominal wall defects, genitourinary malformations and lymphatic or vascular malformations. Each of these has different propensity for syndromic or non-syndromic associated malformations. For instance, anorectal malformation, one of the most common major birth defects encountered in paediatric surgery, has other systemic associations in 28% to 60% [8-10].

Congenital anomalies are responsible for 8% of total neonatal deaths [11] and 24% of deaths in children [4]. The mortality from these birth defects reported from developing countries range from 15% to 52% [7, 9, 12] and as high as 73% in children with associated anomalies in other organ-systems [9]. The outcome, although improving in developed countries [13], remains poor in developing nations. This is due largely to such factors as poor awareness of congenital anomalies by women of reproductive age group [14]; absence/suboptimal uptake of formal preventive programmes such as periconceptional folate supplementation/fortification [15]; absence of perinatal screening for fetal congenital anomalies and sub-optimal access to maternal and child health care [4, 6, 8].

Although no known data-driven scientific report in this respect is available, high morbidity and mortality are the oft-reported outcome of care of these birth defects in developing nations. Most studies on this area of care have not provided information on the specific mortality rate in patients who had active intervention as well as a comparative outlook in each major group of anomalies following surgery. Furthermore, the lack of multi-disciplinary oriented studies has been a major limitation to obtaining accurate outcome of management considering that most major defects are handled by multiple groups of physicians. We have, recently, initiated a multidisciplinary birth defects group to address this challenge and improve the care of patients with congenital anomalies in our setting. The aim of this study, therefore, was to describe the pattern of such anomalies treated in our general paediatric surgical practice; document associated malformations, and evaluate the in-hospital outcome of treatment by a multidisciplinary team in a single tertiary hospital in Ibadan, Nigeria.

Materials and methods

This was a retrospective review of all cases of congenital anomalies of general paediatric surgical nature that were treated between January 2009 and December 2013 at the University College Hospital, Ibadan, Nigeria.

The University College Hospital is Nigeria's foremost tertiary university teaching hospital located in the Ibadan metropolis, arguably the largest city in sub-Saharan Africa. The hospital has 850 beds and runs specialist services in over 50 departments and units. Paediatric surgical admissions are accrued through four sources: the surgical outpatients, the accidents and emergency wards, neonataology wards and the children's emergency ward. The hospital is equipped with 12 modular theatre suites and has dedicated paediatric anaesthetists, hence offers surgery under general anesthesia to neonates and children of any age. Specialists in cardiovascular and thoracic surgery, neurosurgery, ophthalmology, oral and maxillofacial surgery, orthopaedics and trauma surgery, otorhinolaryngology, paediatric surgery, plastic and reconstructive surgery and urology perform surgical care for children in the hospital. This study focuses on only the patients operated in general paediatric surgery.

Information retrieved from the charts and operative records of patients admitted to the general paediatric surgical service of the hospital included sociodemographic details (age, gender, educational status and occupations of the parents), some obstetric

details, and family history of birth defects. The specific types of anomalies and other-system associations in each case were recorded. The type of surgery performed and in-hospital outcome of treatment were also retrieved. Patients who had minor anomalies that often do not require surgical treatment (like skin tags, small haemangiomas, isolated polydactyly etc) were not captured and were excluded from the study.

Results

During the study period 1,539 patients were operated in the division, of which 540 (35.1%) had congenital anomalies. These 540 patients also represented 47.1% of the patients in the larger multidisciplinary birth defects pool. The majority, 419 (77.6%), were males. The ages of the patients ranged from first day of life to 23 years with a median of 17.0 months (an outlier was a 23 year old female with anorectal

Table 1: Types of malformations seen over the period grouped according to anatomical/embryological relationship

Group of malformation	Number (%) N = 568	% Male	Median age	Test statistic*	p value
Respiratory (Tracheoesophageal fistula)	11 (1.9)	57.1	22 hours	177.23	< 0.001
Upper GI (Pyloric stenosis, duodenal atresia, jejunoileal atresia, malrotation, biliary atresia)	32 (5.6)	63.0	2.6 months		
Lower GI (Anorectal malformation, Hirschsprung disease)	66 (11.6)	68.3	3 weeks		
Anterior abdominal wall (Omphalocoele, gastroschisis)	29 (5.1)	56.5	24 hours		
Hernia and hydroceles (Inguinal hernia, umbilical hernia, hydrocele)	276 (48.6)	88.0	3 years		
Genitourinary (Posterior urethral valve, hypospadias, undescended testis)	110 (19.4)	94.5	2 years		
Others (Thyroglossal cyst, DSD, sacrococcygeal teratoma, cystic hygroma)	44 (7.7)	54.5	4 months		

NB – Some patients had congenital malformations involving more than one major system
*DAMA – discharge against medical advice, GI – gastrointestinal, DSD – disorder of sexual differentiation. *- Kruskal Wallis Test*

Data were computed and analyzed using the IBM® SPSS version 21 (SPSS Inc, IBM Corp, Armonk-NY, USA). Descriptive variables were summarized using means and standard deviations (or median and range) for continuous data, and frequencies, ratios and proportions for categorical data. For the purposes of bivariate analysis, variables were dichotomized according to defined responses. In-hospital outcome was classified as alive, discharged against medical advice or dead during in-patient stay. Tests of association between variables were conducted using Chi-square statistics or likelihood ratio and medians were compared using the Kruskal Wallis test. The p-value for statistical significance was set at < 0.05.

malformation – the other patients age ranged from the first day of life to 15 years). The median ages at presentation were lowest in patients with respiratory anomalies (22 hours) and anterior abdominal wall defects (24 hours) and highest in those with hernias and hydroceles (3 years) and genitourinary anomalies (2 years), $p < 0.001$ (Table 1). A total of 223 (41.3%) patients presented within the first year of life. Twelve (2.2%) patients had twin brothers or sisters. The median age of the patients' mothers was 31.6 years, range (18 to 49 years). There was no record of consanguineous relationships.

The most common major congenital lesions documented in this retrospective database were: anorectal malformations in 51 (9.4%), exomphalos

Table 2: Associated major congenital malformations seen in the patients

Group of malformation	Number with major malformations in other systems (%)	Systems involved
Respiratory	7 (63.6)	Congenital Heart Disease (3), Upper GI (1), Lower GI (1), Genitourinary (2)
Upper GI	5 (15.6)	Respiratory (1), Genitourinary (3), Anterior abdominal wall defect (1)
Lower GI	16 (24.2)	Respiratory (1), Cleft lip/palate (2), Genitourinary (10), Limb malformations (3)
Anterior abdominal wall	8 (27.6)	Congenital Heart Disease (3), Cleft lip/palate (2), Upper GI (1), CNS (1), Limb malformations (1)
Hernia and hydroceles	29 (10.5)	Congenital Heart Disease (1), Upper GI (3), CNS (3), Genitourinary (18), Limb malformations (2), Anterior abdominal wall defect (2)
Genitourinary	26 (23.6)	Congenital Heart Disease (5), Respiratory (2), Upper GI (3), Lower GI (10), Cleft lip/palate (2), CNS (4)
Others	7 (15.9)	Congenital Heart Disease (4), CNS (3)

CNS = central nervous system; GI = gastrointestinal

Table 3: Outcome of treatment of patients with congenital malformations

Group of malformation	Died (%)	DAMA (%)	Alive (%)	Unknown (%)	Total (%) N = 568
Respiratory	7 (63.6)	1 (9.1)	3 (27.3)	0	11 (1.9)
Upper GI	4 (12.5)	1 (3.1)	25 (78.1)	2 (6.3)	32 (5.6)
Lower GI	2 (3.0)	0	62 (93.9)	2 (3.0)	66 (11.6)
Anterior abdominal wall	4 (13.8)	1 (3.4)	22 (75.9)	2 (6.9)	29 (5.1)
Hernia and hydroceles	1 (0.4)	1 (0.4)	268 (97.1)	6 (2.2)	276 (48.6)
Genitourinary	0	0	103 (93.6)	7 (6.4)	110 (19.4)
Others	1 (2.3)	2 (4.5)	37 (84.1)	4 (9.1)	44 (7.7)

DAMA – discharge against medical advice, GI – gastrointestinal, Unknown – implies missing data on survival status (Likelihood ratio statistic = 78.375, $p < 0.001$).

Table 4: Age at presentation vs. in-hospital outcome of treatment

Age at presentation	Alive (%)	Died (%)	DAMA (%)	Total (%) N = 518*	χ^2	p value
< 28 days	101 (83.5)	17 (14.0)	3 (2.5)	121 (100.0)	49.906	< 0.001
4 weeks – 1 year	112 (96.6)	1 (0.9)	3 (2.6)	116 (100.0)		
> 1 year	280 (99.6)	1 (0.4)	0 (0.0)	281 (100.0)		
Total	493 (95.2)	19 (3.7)	6 (1.1)	518 (100.0)		

*DAMA – Discharged against medical advice, * – 22 patients had incomplete records*

in 27 (5.0%), hypospadias in 27 (5.0%), Hirschsprung disease in 15 (2.8%), posterior urethral valve in 15 (2.8%) and tracheoesophageal fistula in 11 (2.0%) patients. Table 1 shows the distribution of the types of anomalies in each major grouping.

Associated major lesions were most prevalent amongst respiratory (63.6%), anterior abdominal wall (27.6%), lower gastrointestinal (24.2%) and genitourinary (23.6%) organ systems. Anomalies of the genitourinary system were seen to have occurred

in association with virtually all the other systemic groups of congenital defects respectively (Table 2).

A total of 19 (3.5%) patients died during their primary hospital admission, 6 (1.1%) were discharged against medical advice, 22 (4.1%) had incomplete records on survival status and 493 (91.3%) were alive as at the time of discharge or last follow up visit. Postoperative mortality was highest in patients with tracheoesophageal fistula and lowest in those with genitourinary anomalies, $p < 0.001$ (Table 3). The proportion of patients who died amongst those who presented within the first four weeks of life was higher than those who died after presentation at older ages, $p < 0.001$ (Table 4).

Discussion

Children with congenital anomalies accounted for one-third of all the patients operated in our division of paediatric surgery over the study period. This proportion is within the 24% to 96% reported in the literature [4, 5, 7]. Congenital anomalies account for a significant workload for general paediatric surgeons in both developed and developing countries since most patients present at or shortly after birth. This finding, therefore, perhaps corroborates the fact that birth defects may be as important as other considerations such as infectious diseases in the burden of neonatal health care in developing countries [16, 17].

The patients in this study presented to the hospital late at a median duration of 17 months, and nearly 60% presented outside the first year of life. Patients with more lethal anomalies such as tracheoesophageal fistulas and more grossly apparent defects such as omphalocoeles and gastroschisis presented significantly much earlier than those with other types of defects. In the same vein, patients with non-life threatening anomalies but are likely to have longer lasting physiologic alterations such as patients with genitourinary malformations presented to pediatric surgery quite late in this study. These findings are similar to those from the report of authors working in environments with comparable socioeconomic conditions. Bickler and Sanno-Duanda [4] reported that three quarters of the patients with congenital anomalies presented past infancy in Banjul, the Gambia. Delay in presentation is likely to be multifactorial. A contributory factor may be the cultural influence on the care of children with congenital malformations in the sub-region. This can range from pre-hospital intracommunal consultations to determine the cultural origins of any infantile dysmorphology, to initial attempts at self-help with alternative medicine, and to outright infanticide of children with gross structural birth defects [8, 12].

Nevertheless, delay in recognition both by those who took delivery of the baby as well as by the parents appear to play major roles since many of the malformations e.g. anorectal malformations should be apparent on thorough physical examination of the newborn. Yet as much as 86% of babies with anorectal malformations in Ile-Ife, a nearby town, also presented late to the hospital [8]. Compounding the problem is that 62% of births in Nigeria take place at home with rather little immediate postnatal check up by a registered health care worker [18].

Anorectal malformations, exomphalos and hypospadias were the most common major congenital anomalies treated in the division during the period covered by this study. In terms of the organ systems affected, the gastrointestinal tract accounted for 17% of the malformation in the present study. Kouame *et al.* [12] in a retrospective study conducted at three major teaching hospitals in Cote d'Ivoire over a period of 11 years reported that gastro intestinal anomalies accounted for 13% of the birth defects in their patients and anorectal malformation was the most prevalent in that group. Similarly, Bickler and Sanno-Duanda [4] reported that lower gastrointestinal malformations accounted for the highest proportion, 12%, of major congenital anomalies in the Gambia. Jangra et al. [5] on the other hand reported that 51% of the cases of birth defects in their study conducted in India were due to gastrointestinal malformations and one half of those patients had anorectal malformations. Gastro intestinal malformations are so relatively common likely because of the complex development of the foregut, midgut and hindgut derivatives, which involves interplay between various endodermal and mesodermal elements with different levels of molecular regulations that may ultimately fail [1].

The most critical phase for the development of congenital anomalies is in the first eight weeks of gestation and most of the anlage structures of the gastrointestinal, respiratory and genitourinary systems are formed, in major parts, between the third and eight weeks. Teratogenic insults at this period are likely to affect more than one organ system, thus giving rise to malformations in more than one system. In the present study, the patients had associated lesions in 10.5% to 63.6% of cases with the organ-systems most likely affected by multiple anomalies being the respiratory system, anterior abdominal wall, lower gastrointestinal tract and genitourinary system. These are regions whose embryological

developments are closely linked, hence are likely to suffer from similar interference with their formation.

In-hospital mortality was highest in patients with defects in the respiratory system, anterior abdominal wall and upper gastrointestinal tract. Patients with anomalies in these systems are prone to having multi-system involvement especially congenital heart diseases and other often-rapidly fatal defects. As much as 70% of mortality after surgery in patients with birth defects is attributable to the presence of multiple anomalies [9]. The odd exception in this category, patients with lower gastrointestinal malformations with mortality after surgery of 3%, is not unexpected because the majority of associated defects in patients with anorectal malformations in this study occurred in the genitourinary system. Genitourinary system anomalies were associated with the most favourable outcome in the present study.

A major limitation of this study is its retrospective nature with complete data not always available for the patients. It is a major drawback of a paper-based recording system and the survival status of some of the patients at the end of the study could not be accounted for. Furthermore, this study is tertiary university hospital based and there may be a referral bias in the type of cases that were treated. This calls for the establishment of prospective registry of birth defects in our unit, and ultimately possibly a surveillance system for studying this subject more comprehensively.

In conclusion, patients with birth defects accounted for 35% of the operative workload of this general paediatric surgery unit, and for 47% of all children with congenital anomalies in a multidisciplinary birth defect database in Ibadan, Nigeria. The major congenital anomalies seen were chiefly in the gastrointestinal and genitourinary systems; most patients presented late for surgery and outcome was worst among patients with tracheoesophageal fistula. In-hospital mortality was poorer in patients who had more grossly apparent or symptomatic anomalies and presented earlier.

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