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Congenital craniofacial anomalies: The experience of a sub-Saharan African tertiary hospital.

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Abstract

Background: Congenital craniofacial anomalies range from a simple notch to grotesque craniofacial morphology, which may not be compatible with life. There is dearth of literature on the clinical profile of congenital craniofacial anomalies as an entity in our practice setting in sub-Saharan Africa.

Methods: This study retrospectively analyzed the in-hospital clinical records of individuals with craniofacial anomalies during a five-year period in a foremost university teaching hospital in Nigeria. The information retrieved included the biodata of patients and their parents; the types of cranial-facial anomalies whether isolated or associated with other-system birth defects in each case, treatment received, and the final in-hospital disposition – whether dead or discharged home alive.

Results: There were 200 patients with 272 individual craniofacial anomalies constituting 17.4% of all congenital anomalies in our multidisciplinary birth defect study group database. The median age of presentation was 1.7 months; the craniofacial anomalies occurred in isolation in 77.0% of the cases, and craniofacial clefts were the commonest. The cardiovascular, central nervous and musculoskeletal systems were the most common associated with other-system anomalies. The hospital exit status was good in 96% of those with isolated anomalies compared to the 83% in those with concurrent multiple lesions. Surgical treatment was carried out in 56% of the patients with craniofacial anomalies during the study period.

Conclusion: Craniofacial congenital anomalies represented a substantial proportion of all congenital anomalies seen at our centre. Orofacial clefts were the commonest of these anomalies, majority occurring in isolation and significant proportions of these were amendable to surgical operative intervention.

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Keywords: *Craniofacial, congenital, anomalies, multidisciplinary, in-hospital profile, developing countries*

Résumé

Contexte: Les anomalies craniofaciales congénitales vont d'une entaille simple à une morphologie craniofaciale grotesque, qui peut ne pas être compatible avec la vie. Il existe une pénurie de littérature sur le profil clinique des anomalies craniofaciales congénitales en tant qu'entité dans notre milieu de pratique en Afrique subsaharienne.

Méthodes: Cette étude a rétrospectivement analysé les dossiers cliniques hospitaliers de personnes atteintes d'anomalies craniofaciales pendant une période de cinq ans dans un important hôpital d'enseignement universitaire au Nigéria. Les informations recueillies comprenaient les données biographiques des patients et de leurs parents; les types d'anomalies cranio-faciales isolées ou associées à d'autres anomalies congénitales dans chaque cas, le traitement reçu et la disposition finale dans l'hôpital - qu'ils soient morts ou déchargés chez eux en vie.

Résultats: il y avait 200 patients avec 272 anomalies craniofaciales individuelles constituant 17,4% de toutes les anomalies congénitales dans notre base de données multidisciplinaire de groupe d'étude sur les anomalies de naissance. L'âge médian de présentation était de 1,7 mois; Les anomalies craniofaciales se sont produites isolément dans 77,0% des cas, et les fentes craniofaciales étaient les plus fréquentes. Les systèmes cardiovasculaires, nerveux central et musculo-squelette étaient les anomalies les plus fréquentes associées à d'autres systèmes. Le statut de sortie de l'hôpital était bon chez 96% de ceux atteints d'anomalies isolées comparativement à 83% chez ceux atteints de lésions multiples simultanées. Le traitement chirurgical a été effectué chez 56% des patients atteints d'anomalies craniofaciales pendant la période d'étude.

Conclusion: Les anomalies congénitales craniofaciales représentaient une proportion importante de toutes les anomalies congénitales observées dans notre centre. Les fissures orofaciales étaient les plus fréquentes de ces anomalies, la majorité s'étant isolée et des proportions importantes de celles-ci étaient modifiables pour l'intervention chirurgicale.

Mots-clés: *Craniofacial, congénital, anomalies, multidisciplinaire, profil hospitalier; pays en voie de développement*

Introduction

Congenital craniofacial anomalies are abnormalities of structure and/function that involve the cranium as well as the soft tissues and bones of the face. They include defects like cleft lip, cleft lip and palate, cleft palate, atypical facial clefts, eyelid defects, craniosynostosis, first and second branchial arch defects, mandibular defects and oral defects [1-3]. Their severity ranges from minor affectations such as alopetic defect in the eyebrow, minor notching of the upper eyelid, labial pits, bifid uvula, to grotesque craniofacial disfigurements and anencephalic conditions, which may not be compatible with life [4,5].

Orofacial clefts, have worldwide prevalence rates of between 1 in 700 and 4 in 1000 births with racial and ethnic variations [6-8]. Generally, the prevalence is highest in Asian population (2.4 per 1000 births) followed by the Caucasians, (0.91 to 2.69 per 1000 births) and appears to be lowest in native Africans [6-10]. It is, however, not clear if these differences are the results of under-reporting in the less developed countries. This possibility is buttressed by the general lack of functional and dynamic birth registries in these developing populations. It has also been attributed to the non-uniformity in the classification of craniofacial clefts by different studies [6].

Cleft lip and/palate are the commonest craniofacial clefts reported in global literature [6]. They occur in isolation in about 70% of cases or as components of recognized congenital syndromes such as Van der Woude, Pierre Robins and Treacher Collins [6].

The burden of these anomalies is especially not well documented in developing countries. In an effort to investigate and possibly manage these anomalies the need for a dynamic surveillance programme is imperative. However, a baseline data is an integral requirement for setting up a surveillance programme [11]. Thus, it is the aim of this study to provide a multi-disciplinary baseline data for congenital craniofacial anomalies from the premier university teaching hospital in Nigeria.

Materials and methods

This study was a five-year cross-sectional review of cases seen between January 2009 and December 2013 at the University College Hospital, Ibadan, Nigeria. Patients with major structural congenital craniofacial anomalies managed in this hospital over the study period were included. Cases of congenital craniofacial anomalies were extracted from a larger pool of the multidisciplinary data-set of our institution's birth defect study group. These birth defect data-sets were from the hospital's paper-based records. Congenital craniofacial anomaly was defined as any structural craniofacial abnormality present at birth. They were recruited from the records of the managing specialty units including the paediatric surgery, neurosurgery and the orofacial cleft units. Congenital craniofacial anomalies were grouped into five broad types: craniofacial clefts, congenital hydrocephalus, encephalocoele, craniosynostosis and microcephaly. Craniofacial clefts consisted of four subtypes namely; cleft lip alone, cleft palate alone, cleft lip and palate and rare craniofacial clefts.

Case notes were retrieved and patients' data were extracted and managed via an initial dual data entry using epidata version 3.1 and analyzed with IBM® SPSS version 21. Duplicate entry of patient information into the database was prevented using the SPSS software to detect identical hospital numbers and names. Information on biodata, types of craniofacial anomalies, associated anomalies, surgical intervention and hospital exit status was obtained using a proforma on birth defects predesigned to record the targeted study variables. Hospital exit status was determined by the condition of the patients at the time of discharge (discharged alive, dead, and discharged against medical advice - DAMA). Discharged-alive was considered a satisfactory hospital exit status while dead or DAMA was considered unsatisfactory.

The Chi-square test was used to determine the effect of categorical variables such as surgical intervention, gender distribution and occurrence of multiple associated anomalies on hospital exit status. The level of statistical significance was placed at $p<0.05$.

Results

A total of 200 patients presented with 272 congenital craniofacial anomalies over the 5-year period reviewed. These craniofacial anomalies represented

Table 1: Biodata of patients with congenital craniofacial anomalies

Patient age			
•Mean = 18.2 months (SD+/-48.3) •<1 year - 152 (76.0%) •1 - 10 years - 40 (20.0%) •>10 years - 8 (4.0%)		Median = 1.7 months	Age range = 1 hour to 35 years
Gender distribution	Male - 89 (44.5%)	Female - 100 (50.0%)	
Mothers' age	Mean = 31.9 years (SD+/-6.2)	Median = 31 years	Age range= 19 - 52 years

Table 2: Table of Craniofacial anomalies

Anomalies (No. of patients)	Gender M=Male F=Female	Number with Isolated anomaly (%)	Number with more than 1 anomaly(%)	Surgery (%)	Outcome (%)
Cleft lip alone (74)	M = 33 F = 41	71 (95.9)	3 (4.1)	Y= 45 (60.8)* N= 28 (37.8)	Discharged= 73 (98.6)* DAMA= 0 Died= 0
Cleft palate alone (25)	M =10 F = 15	23 (92.0)	2 (8.0)	Y= 12 (48.0)* N= 11 (44.0)	Discharged= 24 (96.0)* DAMA= 0 Died= 0
Cleft lip and palate (16)	M = 7* F = 8	11 (68.8)	5 (31.3)	Y= 10 (62.5)* N= 5 (31.3)	Discharged = 14 (87.5) DAMA= 2 (12.5) Died= 0
Rare craniofacial clefts (4)	M = 3 F = 1	1 (25.0)	3 (75.0)	Y= 2 (50.0) N= 2 (50.0)	Discharged = 2* DAMA=0 Died=0
Hydrocephalus (60)	M = 28* F = 23	47 (78.3)	13 (21.7)	Y= 35* N= 19	Discharged = 46* DAMA= 3 Died= 3
Cranial encephalocoele (8)	M = 3 F = 5	6 (75.0)	2 (25.0)	Y= 7* N=0	Discharged =6 DAMA=1 Died=1
Craniosynostosis (3)	M = 2 F = 1	3	0	Y= 0 N= 3	Discharged = 2* DAMA=0 Died=0
Microcephaly (14)	M =5* F = 8	3 (21.4)	11 (78.6)	Y=4* N=9	Discharged =9* DAMA=3 Died= 0

* Do not add up because of missing data.

17.4% of all birth defects encountered at the hospital during the period. There were 89 males and the mean age was 18.2 months. Their biodata are as presented in table 1. Craniofacial clefts accounted for the majority of the cases while the craniosynostosis was the least common (Table 2). Isolated craniofacial anomaly occurred in 77.0 % of the cases. Microcephaly had the highest proportion of associated other-system anomalies (Table 2). The associated anomalies were found in similar proportions in the cardiovascular system, central nervous system, orbital region, and musculoskeletal system. The details of the other-system anomalies are as shown in table 3.

Fifty-six percent of the patients had surgical intervention for their anomalies; 87.0% were discharged alive; 2.0% died and 4.5% obtained discharge against medical advice. For the purpose of statistical analysis, the 'Type of craniofacial anomaly' was re-categorized as craniofacial clefts and other craniofacial anomalies (Table 4). The craniofacial cleft group all had satisfactory in-hospital outcome while about a seventh of the other

Table 3: Types and frequency of associated other system anomalies

	CVS	GIT	CNS	EG	GUT	MSS	CI	Eye	ENT	AAWD	V
Cleft lip anomaly 74	1- VSD	_	1-H	1-DS	-	1-A	-	-	-	-	-
Cleft palate anomaly 25	1- VSD	-	-	1-DS	1-UT	-	-	1-WG	-	-	-
Cleft lip and palate anomaly16	1- VSD	-	1-M	-	-	1-P 1-HT	-	1-OA	-	1-OMP	-
Rare clefts 4	1- VSD	-	1-CE	1-PS	1- Hp	-	-	-	1-AA	-	-
Hydrocephalus 60	1- VSD	1-CL	7-SB 1-CE	-	1-Hp 1-UT	3-T 1-S	-	-	-	1-IH	1- CH
Microcephaly 14	4-VSD 2-PDA	1-CLP	1-SB	1-DS	2-KM 1-Hp 1-UT	1-T 2-P	5-CRS	6-CA 1-WG 1-CO	1-M	1-IH	-
Cranial encephalocoele 8	-	1- RC	-	1-PS	-	-	-	-	-	-	-
Total 204	11	3	11	5	7	11	5	10	2	3	1

RT- Respiratory tract, CVS- Cardiovascular system, CNS- Central nervous system, EG- Endocrine Genetic, GUT-Genitourinary tract, MSS- Musculoskeletal system, CI-Congenital infections, ENT-Ear Nose and Throat, AAWD- Anterior abdominal wall defect, V- Vascular, CL- Cleft lip, CP- Cleft palate, CLP-Cleft lip and palate, VSD Ventricular sepatal defect, PDA-Patent ductus arteriosus, DS- Down syndrome, PS-Patau syndrome, UT-Undescended testes, CE- Cranial encephalocoele, H- Hydrocephalus, M-Microcephaly, SB- Spina bifida, RC-Rare craniofacial clefts, Hp-Hypospadias, KM- Kidney malformation, A- Achondroplasia, P- Polydactyly, HT-Hypoplastic thumb, T- Tallipes, S-Syndactyly, CRS- Congenital rubella syndrome, WG- Whole globe abnormalities, OA- Orbital abnormalities, CA- Cataract, CO- Congenital corneal opacity, AA- Aural atresia, M- Microtia, OMP- omphalocoele, IH- Inguinal hernia, CH- Cystic hygroma.

Table 4: Table of associations

		Hospital Exit Status			
		Discharged (%)	Dead (%)	DAMA (%)	p-value
Type of craniofacial anomaly	Craniofacial clefts	100 (100.0)	0 (0.0)	0 (0.0)	<0.005
	Other craniofacial anomalies	60 (84.5)	4 (5.6)	7 (9.9)	
Gender	Male	76 (95.0)	2 (2.5)	2 (2.5)	0.099
	Female	88 (93.6)	0 (0.0)	6 (6.4)	
Number of anomalies	Single	149 (95.5)	4 (2.6)	3 (1.9)	0.005
	Multiple	24 (82.8)	0 (0.0)	5 (17.2)	

Note:
Craniofacial clefts = cleft lip alone + cleft palate alone + cleft lip and palate +other craniofacial clefts
Other craniofacial anomalies = congenital hydrocephalus + microcephaly + cranial encephalocoele + craniosynostosis

craniofacial anomalies (15.5%) had unsatisfactory in-hospital outcome. In addition, only a minority (4.5%) of the patients with isolated (single) anomaly had unsatisfactory hospital exit status while 17.2% of patients with multiple anomalies had unsatisfactory hospital exit status, majority being discharged against medical advice. The difference was statistically significant (Table 4).

Discussion

Craniofacial anomalies are a varied group of birth defects seldom reported as a whole. In the literature, the term 'craniofacial anomalies' is often used mainly in reference to craniofacial clefts. Sometimes, however, it is also used to capture cases of congenital hydrocephalus, craniosynostosis, and encephalocoele which occur either in isolation or as components of neural tube defects (NTDs), and which may not be limited to the craniofacial region [12,13]. It was therefore difficult to compare the prevalence rate in this study to most reports in the literature. In this study, the prevalence rate is 17.4% which is slightly lower than previous African reports of 20.8% and 24.5% in Nigeria and Abidjan respectively [14,15].

Craniofacial clefts were the commonest craniofacial anomalies observed in this study. This is similar to findings previously documented by other studies [2, 3,16-18]. Cleft of the lip alone (CL) and cleft of the lip and palate (CLP) have been considered to be variants of the same entity but of varying severity [8]. Therefore, they are generally referred to as cleft of the lip and/or palate (CL/P). However, there is yet other evidence that CL and CLP may not be variants of same entity, as attempts have been made to demonstrate the differences between these two anomalies and there are publications that reported them separately [3,7,18,19]. They are therefore considered separately in this study for the purpose of clarity. Cleft Lip was the most common of all craniofacial anomalies as well as among all craniofacial clefts. Contradictory reports of the type of craniofacial clefts with the highest frequency exist in the literature. From their study, Kesande *et al* reported CL as the commonest while others like Aziza *et al* reported CP as the commonest [3,7]. Craniosynostosis, which has been reported as one of the commonest of the cranial anomalies occurring as 1 in 2,000 to 2,500 live births was the least represented in our series [3-20]. Anencephaly and spina bifida are regarded as the most common of the neural tube defects [5]. No case of anencephaly was recorded in our study. The retrospective nature of our study as well as the ward admission-based acquisition of the data from the neurosurgical unit may account for these observed dissimilarities with literature reports since cases of craniosynostosis not admitted on the wards were not captured in our data. The lack of anencephalic cases (a condition that is not compatible with life) may be due to the unavailability of stillbirth records in our centre during the period of this study.

In our study, the maternal age range was 25 to 35 years, a very young maternal population indeed. This was similar to the findings of Onankpa in Sokoto in Nigeria and Kesande in Uganda who reported maternal mean age of 26 years and 55.0% of the mothers younger than 30 years respectively [7,14].

Craniofacial anomalies, especially orofacial clefts, have been reported to be more common in males than females [3,7]. While CL/P is said to be

more prevalent in males, CP is commoner in females in some studies; still, other reports revealed no gender difference [8,16,19,21-23]. Very few studies have reported female preponderance for CL/P [6,7]. In our study, there were more females than males with craniofacial anomalies. The authors do not know the reason for this, apart from the possible bias of the retrospective nature of this analysis.

Our study revealed that the occurrence of both non-cleft congenital craniofacial anomaly and multiple congenital anomalies both significantly predict an unsatisfactory in-hospital outcome (Table 4). The reason for the poor outcome needs to be investigated; however, financial constraints, poor health facilities and lack of expertise to manage these complicated cases are possible reasons.

Distinguishing between the isolated and multiple anomaly cases may shed some light on the nature of aetiology of these anomalies [2]. In this study majority of the craniofacial clefts occurred as isolated clefts. However, the proportion of cases occurring with associated anomalies (10.9%) is, on the one hand, higher than that reported by Butali *et al* [20] who documented a rate of 4.7%; and, on the other hand, lower than 30% reported by the WHO registry on craniofacial anomalies, as well as the 18% and 50% reported in some other studies [3,24]. The CLP was the most occurring craniofacial cleft with associated anomalies. This is similar to the report of Jugessur *et al* [18] in Norway but differs from the documentation of Marazita [7], which stated that CP alone has a higher percentage of associated anomalies [7,18]. The cardiovascular system anomaly was the one with the most prevalent association with congenital craniofacial anomalies. This was similar to the findings in literature [2,3].

Increasing number of congenital craniofacial anomalies, especially the craniofacial clefts, are benefitting from surgical correction and therefore finding some sort of solution. Nevertheless, affected patients and their parents or caregivers may have to endure multiple and expensive interventions practically throughout an affected individual's life time with attendant psychological effects. A means of prevention will no doubt be a better and cheaper option. However, for this option to be a reality, these anomalies need to be understood. The first step in understanding them will require collection of representative data, which can be obtained adequately through prospective birth defect surveillance programmes as proposed by WHO in 2010 [25]. This further emphasizes the need to set up birth defect surveillance in our environment and in Africa as a whole [11].

This study demonstrates the benefits of a multidisciplinary approach to research. It is the first of its kind in our centre to look at congenital craniofacial anomalies as a whole as well as their association with other system anomalies. However, this being a retrospective study was challenged by shortcomings such as missing data. A more comprehensive prevalence study will require additional information on prenatal screening, terminated pregnancies and stillbirths in order to appreciate the true magnitude of congenital craniofacial anomalies' burden in our environment and to compare with similar data from the registries of industrialized countries.

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