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PATTERN OF OROFACIAL CLEFTS: A STUDY OF 673 CASES IN NORTHCENTRAL NIGERIA

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Abstract

Background: Orofacial cleft presentation has been shown to have geographical variation. The objective of this study is to determine the pattern of orofacial cleft presentation in Northcentral, Nigeria.

Materials and Methods: This is a study of orofacial cleft patients seen in the Nigerian Northcentral zone from 2011 to 2017, at University of Abuja Teaching Hospital as well as during outreach programmes by Cleft and Facial Deformity Foundation. Information retrieved from patients' notes included age at presentation, gender, gestation period, maternal history, type of cleft, family cleft history and associated abnormalities. Data collected was analyzed using Statistical Package for the Social Sciences (SPSS) version 23 (SPSS Inc, Chicago IL, USA).

Results: A total of 673 patients were included in the study. Cleft ratio recorded was Cleft Lip (CL); 55.6%, Cleft lip & Palate (CLP); 19.5% and Cleft Palate (CP); 12.6 There were 85 unilateral complete cleft palate (UCLP) and 43 bilateral cleft lip and palate (BCLP) with ratio UCLP: BCLP of 2:1. Unilateral cleft lip was higher than bilateral cleft lip in the ratio of 8.7:1. Forty-four (6.5%) patients had a famililial history of cleft lip and/or palate occurence, 4.2% had associated abnormalities and 5.3% had atypical facial clefts.

Conclusion: Pattern of cleft presentation is similar to earlier reports, including adult cleft. Significant findings included high percentage of isolated cleft palate as well as unilateral cleft lip being nine times commoner than bilateral cleft lip. Awareness creation and access to surgery offered by outreach programs have the potential of reducing adult cleft.

Introduction

"Whenever a lip, in particular the uppermost one, is cleft by birth or nature, it is called a harelip, because in hares the uppermost lip is quasi-cleft too. It causes great ugliness of the face but if the cleft is large, such people cannot help speaking nastily and unacceptably through their nose for the term of their life." —Lorenz Heister, 18th Century German Anatomist and Surgeon[1]

Orofacial clefts (OFCs) are common congenital malformations whose etiology is complex and likely to have both genetic and environmental underpinnings.[2,3] It could affect the lip, the palate and rarely any other part of the face. OFC could be isolated and it could be syndromic (found in association with some other systemic abnormalities).[4]

The pattern of orofacial cleft presentation has been shown to have geographical and ethnic variation.[5]

The reviewed studies on the birth prevalence of orofacial clefts in Africa suggest that the prevalence is low compared to Europe, America, and Asia.[5] In fact, racial comparative analysis revealed a cleft occurrence of one in every 300 live births in native Americans, one in 2500 African live births and one in every 600-700 live births in Asian and European populations.[6] Birth prevalence of OFC from different African populations vary widely, from as low as 0.3/1,000 reported in Nigeria [7] to 1.65/1,000 reported in Kenya.[8] The difference in prevalence across the world provides support for the role of multiple genetic and environmental factors that increases the risk for OFCs. OFCs occur following disturbances of the normal genomic architecture during facial development, an environmental insult around this time increases the risk for OFCs.[9]

The surgical literature is consistent in recommending early repair of orofacial cleft, typically completing primary repair of the lip and palate by the age of two years to prevent psychological trauma in these individuals with cleft lip and/or palate. While this is the case in the developed world, lack of access to care or surgery and poverty make early repair difficult and as such late presentation is common in Africa and India subcontinents.[10,11] Free surgical mission is a common phenomenon in developing countries which typically was carried out by specialists from developed countries.[10,11,12] In recent times, local initiatives are being reported in the literature.[13]

Northcentral Nigeria is one of the six geopolitical zones of the sub-Saharan African nation. The zone is sub-divided into seven provinces including the Federal Capital Territory. Two of the provinces (Kwara and Plateau) have specialized health facilities where cleft repairs are undertaken while the other provinces (Benue, Kogi, Nassarawa, Niger and FCT with a population of 14,7 million[14] depend mainly on a facility located in Abuja (University of Abuja Teaching Hospital [UATH]) and an indigenous free surgical mission (Cleft &Facial Deformity Foundation (CFDF), in partnership with Smile train International, TY Danjuma foundation, Maizube foundation and local philanthropists.

Determining the pattern of presentation of orofacial clefts in a particular area has implications for public health practice, strategies for prevention and treatment .[5] The pattern of orofacial cleft has been presented from different parts of Nigeria,[5,9] but to the best of our knowledge, none has come from the Northcentral region. The objective of this study therefore was to determine the pattern of OFC presentation in the zone.

Material and Methods

This is a retrospective study of patients with OFCs seen between 2011 and 2017. We obtained data from the case notes of patients seen at UATH Abuja and the outreach programmes. CFDF is an indigenous surgical mission outfit, organizing free outreach programmes within the region since 2011. UATH is one of the major hospitals in the northcentral zone where cleft surgery is undertaken. The study was carried out following ethical clearance from the Research and Ethics Committee of UATH, Abuja.

CFDF outreach programmes took place in different hospitals located in all the 5 States (Benue, Kogi, Nassarawa, Niger and FCT), of Northcentral Nigeria. The hospitals include: General hospitals in Kuje, Kwali, Bwari and Karsi towns in the FCT, Specialist hospitals in Lokoja, Minna, Lafia and Keffi towns.

Information retrieved from the case notes included patients' ages at presentation, gender, state/province of origin, gestation period, maternal history of smoking and alcohol consumption, type of cleft, family cleft history and associated abnormalities.

Data collected was analyzed using Statistical Package for the Social Sciences (SPSS) version 23 (SPSS Inc, Chicago IL, USA).

Results

A total of 673 patients were studied, 337 males (50.1%) and 336 (49.9%) females. There were

101 patients from the hospital setting and 572 cases from outreach programmes. The mean (± SD) age of all the patients was 8.1± 10.2 years, that of hospital-based cases was 4.1± 6.9 while that of patients from outreach programmes-based patients was 8.7 ± 11.0 years.

According to the history provided by the patients/informants, 61.2% (n=411) had complete 9-month gestational period; 256 patients (38.1%) claimed no knowledge while 6 patients (0.9%) had gestational periods above 9 months. None of the mothers volunteered positive history of smoking or alcohol consumption. Forty-four patients (6.5%) had a positive family history of OFC, 509 patients (75.6%) had none and 120 patients (17.8%) claimed no knowledge of the occurrence of OFC in their families. Twenty-eight patients (4.2%) had cleft associated abnormalities, while 645 (95.8%) patients had none. Associated abnormalities encountered, based on clinical examination, included facial bone disproportionality, ocular anomalies, polydactyl and syndactyl.

Table 1 shows age distribution of the patients. Three hundred and two patients (44.9%) were aged 2 years and below, 479 patients (71.2%) were less than 10 years, 90 patients (13.4%) were above 20 years while 5 (0.7%) patients were more than 50 years of age.

Table 2 shows the average age distribution of patients recorded per year from 2011 to 2017. The average age of hospital based patients was 4.1± 6.9 and that of outreach programmes was 8.5± 11.0; the difference was statistically significant (p=0.01). The average age recorded varied with each year (12.9±13.2 years in 2011 and 6.1±8.8 years in 2017).

Figure 1 shows the pattern of cleft presentation of the patients. Unilateral cleft lip accounted for 381 (56.7%) cases, bilateral cleft lip 44 (6.5%), isolated cleft palate was 84 (12.5 %) while combined cleft lip and palate was 129 (19.2 %). Right cleft lip accounted for 150 (22.3%) cases while left cleft lip was 267 (34.3 %), with a ratio left: right of 1.8:1.There were 85 unilateral complete cleft palate (UCLP) and 43 bilateral cleft lip and palate (BCLP) with ratio UCLP: BCLP of 2:1.Unilateral cleft lip was higher than bilateral cleft lip in the ratio of 8.7:1. There were 637 (94.7 %) facial clefts {cleft lip and palate}and 36 (5.3 %) atypical cleft (rare clefts). The ratio of CL: 381(56.7%); CLP: 129 (19.2 %); CP: 84 (12.5 %) is shown.

Table 3 shows the gender distribution of orofacial clefts among the patients from Northcentral zone.

A male preponderance was observed in complete lip and palate cases, (M: F of 1.5:1). A female preponderance was reported with cases of isolated cleft palate (M: F, 1:1.5).

Discussion

Aetiology of orofacial cleft is not known but both genetic and environmental factors have been mentioned.[3] In our sample, 6.5% of the patients had a positive family history which is an evidence of genetic aetiology. This is similar to the findings in Northern Nigeria where similar percentages were recorded.[15] Gorlin et al[16] observed that race, ethnicity, geographic locations, environmental factors, and socioeconomic status influence the incidence of OFCs, which presuppose that the relative contributions of individual susceptibility genes may vary across different human populations. The etiology of nonsyndromic orofacial clefts (NSOFCs) is complex, exhibiting multifactorial pattern of inheritance.[17] Earlier Genome-wide Association Studies (GWAS) suggested that the 8q.24 locus may be a risk locus for non-syndromic orofacial clefts across major ethnicities[18,19] Our patients are part of the samples from an African study where for the first time, it was shown that the 8q24 locus is a risk locus in Africans.[17] In a follow-up GWAS, a novel GRHL3 variant was also identified among isolated cleft palate African patients.[20] Gowan et al[17] emphasized the understanding of the genetic architecture of NSOFCs in Africans and further suggests the need to carry out GWASs and whole genome sequencing for every ethnicity as far as complex traits are concerned.

Environmental factors have equally been mentioned as possible etiological factors. None of the mothers of the patients had positive history of smoking and alcohol consumption probably because the sociocultural disposition of the communities in Northcentral Nigeria abhors

Table 1: Age at Presentation of orofacial cleft patients from North Central, Nigeria.

Age Range(years)	Frequency	Percentage
0-2	302	44.9
3-5	85	12.6
6-10	92	13.7
11-15	55	8.2
16-20	49	7.3
21-25	27	4.0
26-30	35	5.2
31-35	11	1.6
36-40	5	0.7
41-45	3	0.4
46-50	4	0.6
>50	5	0.7
Total	673	100.0

Table 3: Pattern of presentation and Gender distribution among orofacial clefts patients from North Central zone

	Hospital Based patients	Outreach programme patients						
	2011-2016	2011	2012	2013	2014	2015	2016	2017
N	101	186	76	97	37	38	42	96
Mean	4.1144	12.8517	8.5529	6.3820	9.3708	8.8079	8.7214	6.1027
Std. Deviation	6.85515	13.22092	10.55822	8.92989	10.42854	11.12134	11.21140	8.86427

women being seen smoking or consuming alcohol in public.

The age of presentation of orofacial cleft patients is affected by many factors such as poverty, inaccessible health care and dearth of specialists. The average age of patients that attended outreach programme was 8.7 years while that of the hospital patients was 4.1 years. Surgical mission programmes in the developing countries are usually characterized by adult attendees who have been variously described as 'adult cleft' or 'unrepaired cleft'.[21,22,23] Since poverty, inaccessible healthcare and dearth of specialists are the common reasons for unrepaired adult clefts, taking surgical access close to the communities encourage better attendance and hence adult age presentation. Conway et al [24] in a review of 36,384 African patients by 389 African surgeons recorded an average age of 9.34 years. The lower age group reported among the patients treated in the hospital setting has also been reported from similar studies from sub-Saharan Africa.[25,26,27] Oginni et al[26] reported over 87% of the patients treated at Ile Ife, southwestern Nigeria were less than 4 years while majority of patients treated in Daresalaam, Tanzania and Nairobi, Kenya were less than 5 years of age.[25,27]

Majority of those attending hospitals usually have some level of education which influenced their early presentation.[26] On the other hand, majority of patients at outreach programmes are essentially uneducated, ignorant and poor and were only persuaded by the free nature and proximity of the surgical service. However our observation revealed a progressive decrease in the age of presentation from 2011 to 2017 with the average age in 2017 being 6 years compared with 12 years in 2011 (Table 4). This implies that improved access to surgical intervention among Nigerians coupled with awareness creation by such programmes provides a hope for the elimination of older unrepaired OFC patients among Nigerians and Africans.

This study reported no sex predilection among the patients, similar to the findings of Adeyemo et al.[2] However some other studies have reported a slight male preponderance.[10,24,25,26,28] This study has recorded a male preponderance among patients with complete cleft lip and palate and a female preponderance among patients with isolated cleft palate. This was equally reported by earlier studies [9,15] In fact, a study by Adeola et al[15] reported a male to female ratio of 14:1 among patients with complete cleft lip and palate from northern part of Nigeria.

The pattern of cleft presentation recorded in this study is comparable to earlier findings. The ratio of CL; 55.6% CLP; 19.5% and CP; 12.6% recorded in this study is not too different from other studies.[15,24] Adeola et al[15], in a review of 500 patients from Northern Nigeria, reported a preponderance of cleft lip compared with cleft lip and palate. Nagalo et al[28] in a review of 185 orofacial cleft patients from Burkina Faso, Sub-Saharan Africa, recorded an almost equal ratio between Cleft lip alone compared with cleft lip and Palate. Conversely, in a review of 36,384 African patients by 389 African surgeons, Conway et al[24] found Cleft lip and palate higher than Cleft lip alone. However all these studies aforementioned reported the percentage contribution of isolated cleft palate as being the lowest with a range of 1.6 to 12.6%. Scientific studies have been consistent in reporting a higher percentage of cleft lip compared with isolated cleft palate and this observation could be considered a scientific standard. The higher percentage of cleft palate patients in our

Table 3: Pattern of presentation and Gender distribution among orofacial clefts patients from North Central zone

Cleft Type	Gender		Total (%)
	Male No (%)	Female No (%)	
Right Incomplete Cleft lip	12(3.6)	8(2.4)	20(3.0)
Left Incomplete Cleft lip	20(5.9)	20(6.0)	40(5.9)
Right Cleft lip	61(18.1)	69(20.5)	130(19.3)
Left Cleft lip	97(28.8)	94(28.0)	191(28.4)
Bilateral cleft lip	21(6.2)	23(6.8)	44(6.5)
Right Cleft Lip and Palate	24(7.1)	16(4.8)	40(5.9)
Left Cleft Lip and Palate	28(8.3)	17(5.1)	45(6.7)
Bilateral Cleft Lip and Palate	24(7.1)	19(5.6)	43(6.4)
Cleft Palate	33(9.8)	51(15.2)	84(12.5)
Atypical Facial Cleft	17(5.1)	19(5.6)	36(5.4)
Total	337(100.0)	336(100.0)	673(100.0)

collection could be due to the nature of the surgical mission as individuals were encouraged to present for surgery due to the free nature of treatment.

In this study, among those who presented cleft lip, left sided cleft lip was mostly observed, this was similarly observed by many studies.[15,24,27-30]

The ratio of unilateral complete cleft lip and palate (UCLP) to bilateral cleft lip and palate (BCLP) recorded in this study was 2:1. A similar but slightly higher finding was made by Butali et al[9] and Conway et al[24] where ratios 2.3:1 and 2.9:1 were observed respectively. This study also recorded a higher prevalence of unilateral cleft lip than bilateral cleft lip with a ratio of 8.7:1. Comparison of observation with other studies is difficult because most studies pay little attention to the ratio.

Reporting associated abnormalities with OFC is confusing, resulting in a wide range of reported figures and this is due to variation in the constituents of abnormalities coupled with lack of routine comprehensive assessment of OFC patients. This study observed that 4.2% of study individuals had associated abnormalities while 5.3% had atypical facial clefts. The prevalence of cleft associated abnormalities reported in our study falls within the wide prevalence rates of 1.9% - 64.2% that have been described in literature.[25,26,31,32] Rawashdeh and Abu-Hawas[3] in a review of 196 Jordanians with OFC found that 14% of patients had associated abnormalities. The affected systems in their study included the cardiovascular system, skeletal system, central nervous system, urogenital system, the eyes, the ears, and digestive system. They advocated routine screening for other malformations in infants with clefts, and genetic counseling in most of these complicated cases. Several other studies paid little attention to comprehensive assessment of cleft patients. While this study documented the prevalence of atypical facial clefts (5.3%), several studies considered them as associated abnormalities while others are silent about it.

Conclusion

Genetic and environmental factors are implicated in the etiology of OFCs, however smoking, alcoholism, drugs and consanguineous marriage are rare in Northcentral Nigeria and are not likely important factors. Unrepaired adult cleft is still common during outreach programmes in Nigeria but patients treated in the hospitals are less than 5 years of age. A ratio of Cleft lip; 55.6% Cleft lip & palate; 19.5% and Cleft palate; 12.6% recorded in this study is not too different from earlier studies. However high percentage contribution of isolated cleft palate recorded is probably due to the incentive of free surgery. Left laterality of cleft lip and ratio of UCLP and BCLP of 2:1 recorded in this study is similar to earlier studies but a ratio of 8.7: 1 recorded for unilateral cleft lip to bilateral cleft lip is an uncommon finding. More males had complete cleft involving the lip and palate while more females had isolated cleft palate. Forty-four (6.5%) patients had a famililial history of cleft lip and/or palate occurrence. Even though reporting of associated abnormalities is a bit complex, this study has observed that 4.2% of study individuals had associated abnormalities based on clinical examination while 5.3% had atypical facial clefts. However awareness creation and access to surgery offered by outreach programmes has reduced adult cleft with a hope of eliminating it.

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Abbreviations

UATH – University of Abuja Teaching Hospital; CFDF – Cleft and Facial Deformity Foundation; CL – Cleft lip; CLP – Cleft lip and palate; CP – Cleft Palate; OFC – Orofacial cleft; OFCs – Orofacial clefts; FCT – Federal Capital Territory; UCLP – Unilateral cleft lip and palate; BCLP – Bilateral cleft lip and palate; — Male; F – Female; NSOFC - Nonsyndromic orofacial clefts; GWAS - Genome-wide Association Studies; GRHL – Grainyhead like transcription factor