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Demographic and clinical profile of craniofacial clefts at Comprehensive Rehabilitation Service in Uganda

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Abstract

Background: The true incidence of Craniofacial cleft (CFC) is unknown because of their scarcity and because of the difficulty in recognizing sometimes subtle physical findings in mild malformations. Craniofacial anomalies in the African population are reported infrequently.

Aim: To contribute to the general literature on rare CFC in Uganda and Africa.

Methods: we conducted a retrospective search of patient data over the period 2005 to May 2017 in *the* unit of plastic surgery of CoRSU (Comprehensive Rehabilitation Service in Uganda) hospital, a tertiary hospital in Uganda. Patient with a diagnosis of CFC were picked out. Sixty-six patient's files with clinical diagnosis of CFC including their clinical photographs were found. Frequency data was generated and a frequency distribution table with the observed data was constructed.

Results: Sex distribution showed no significant difference between male and female (1:1,2);the age on admission ranged from 1 day to 83 years; according to the laterality of the cleft, unilateral CFC (left or right side) are more common than midline clefts (Tessier 0; 14; 0,14;30); however, according to the clinical type, Tessier cleft (TC) 0 is the most common TC in our series and is associated with holoprosencephaly. Fifty percent of CFC in our series are syndromic. TC 7 are common in male and have a bilateral predilection.

Conclusion: CFC are a rare set of malformations for which there is a paucity of literature. There is a need to conduct a study with a larger series including CT-Scan in order to analyze more accurate clinical diagnosis.

Keywords: Craniofacial, Cleft, Tessier, Uganda

Résumé

Contexte: L'incidence réelle de la fente craniofaciale est inconnue en raison de sa rareté et de la difficulté à reconnaître des signes physiques parfois subtils lors de malformations légères. Les anomalies craniofaciales dans la population africaine sont rarement rapportées.

But: Contribuer à la littérature générale sur les fentes craniofaciales rares en Ouganda et en Afrique.

Méthodes: nous avons effectué une recherche rétrospective des données des patients sur la période 2005 à Mai 2017 dans l'unité de chirurgie plastique de l'hôpital CoRSU (Comprehensive Rehabilitation Service in Uganda), un hôpital tertiaire en Ouganda. Les patients avec un diagnostic de fente craniofaciale ont été sélectionnés. Soixante-six dossiers de patients avec un diagnostic clinique de fente craniofaciale, y compris leurs photographies cliniques ont été trouvés. Des données de fréquence ont été générées et des tableaux de distribution de fréquence avec les données observées ont été construits.

Résultats: Les fentes craniofaciales constituent une malformation rare. La distribution par sexe n'a pas montré de différence significative entre les hommes et les femmes (1: 1,2), l'âge à l'admission variait de 1 jour à 83 ans; selon la latéralité de la fente, les fentes craniofaciales unilatérales (côté gauche ou droit) sont plus fréquentes que les fentes médianes (Tessier 0; 14; 0,14; 30); cependant, selon le type clinique de la classification de Tessier, la fente craniofaciale 0 de Tessier est la plus rencontrée dans notre série et est associée à l'holoprosencéphalie. 50% des fentes craniofaciales de notre série sont syndromiques. Les fentes craniofaciale 7 de Tessier sont fréquentes chez les sujets masculins et ont une prédilection bilatérale.

Conclusion: Les fentes craniofaciales constituent une entité rare de malformations pour lesquelles il existe peu de littérature. Il y a un besoin de mener une étude avec une plus grande série comprenant des images tomodensitométriques afin d'analyser avec plus de précision le diagnostic clinique.

Mots-clés: craniofacial, fente, Tessier, Ouganda

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Introduction

Congenital anomalies are a major cause of infant mortality and childhood morbidity, affecting 2-3% of all babies. Approximately 1% of these newborns have syndromes or multiple anomalies; craniofacial anomalies are often a component part [1]. CFC are rare congenital malformations of the cranium and face with deficiencies or excesses of tissue along an anatomic line based on embryologic maldevelopment [2].They are among the most disfiguring of all facial anomalies and have a multitude of clinical presentations with different levels of severity [2, 3, 4]. Despite representing an important segment of craniofacial pathology, CFC are a rare set of malformations for which there is a paucity of literature that is often limited to isolated case reports with little objective long-term follow up [5]. In spite of the modern-day advent of new imaging and new surgical techniques, little has been written on the management of the rare CFC since Tessier's initial classification and many factors limit their optimal management [5]. The incidence of these atypical clefts is unknown but is estimated at 1.4 – 4.9 cases per 100 000 live births, therefore approximately 100 times less frequent than the common clefts [3,4]. Cranio-facial anomalies in the African population are reported infrequently. The implications of such anomalies in the African population are different than in developed countries and should therefore be reported [6]. Most studies in the literature are case studies based on a particular type of Tessier cleft. There is lack of studies reporting a big series of different types of TC. This paper should serve to improve a neglected field in which the literature is bereft of reports of demographic and clinic data on CFC, particularly in Africa.

Table 1. Distribution of patient by Age on admission

Age (months)	Number
<6	10
[6-12]	20
]12-24]	12
]24-60]	15
> 60	9
Total	**66**

Materials and methods

We retrospectively studied data from patient's files over a period of 12 years, from 2005 to May 2017. During this period sixty-six cases of TC patients were seen and treated in the plastic and reconstructive surgery Unit of CoRSU hospital. Data collected included age on admission, sex distribution, location and side, types of CFC, associated syndrome and photographs. No systematic CT-scan was done for these patients.

Tessier classification of CFC was used to classify the clefts (**Fig. 2**). Cases of Holoprosencephaly, Hemifacial microsomia, Goldenhar and Treacher Collins syndrome were also identified. Frequency data was generated and a frequency distribution table with the observed data was constructed.

Table 2. Distribution of patient by sex

Sex	Number
Male	30
Female	36
Total	**66**

Results

Distribution of patient by sex reveals that CFC is slightly more common in female in our setting (30 males vs 36 females); however the difference is not significant (ratio M:F = 1:1.2).

In our series **Table 1** Shows that most of patients (20/66) are admitted between 1 year of age and 2 years included; 24 patients were admitted after 2years of age

Table 3. Distribution of craniofacial cleft by location	
Location of craniofacial clefts	**Number**
Facial cleft	63
Cranial cleft	0
Facial & cranial cleft	3
Total	**66**

The **Table 2** reveals that CFC is slightly more common in female in our setting; however the difference is not significant (ratio M:F = 1:1.2)

Majority of CFC diagnosed clinically are facial cleft without extension to the cranium (63/66).No case of isolated cranial cleft was diagnosed (See **Table 3**).

Table 4. Distribution of craniofacial clefts by laterality	
Side	**Number**
Right	18
Left	13
Midline	25
Bilateral	10
Total	**66**

Majority of CFC are unilateral (31/66) with right side more diagnosed (18/66). However, midline clefts are more common than right side cleft in our series (25/66) (See **Table 4**).

Tessier cleft 0 is the commonest clinical type of CFC (20/66) followed by Tessier cleft 7 (17/66); Tessier cleft 30 was seen in 2/66 patients.(See **Figure 5**)

Tessier clefts 7 are more common in most of the cases (7/17) (See Table 7)

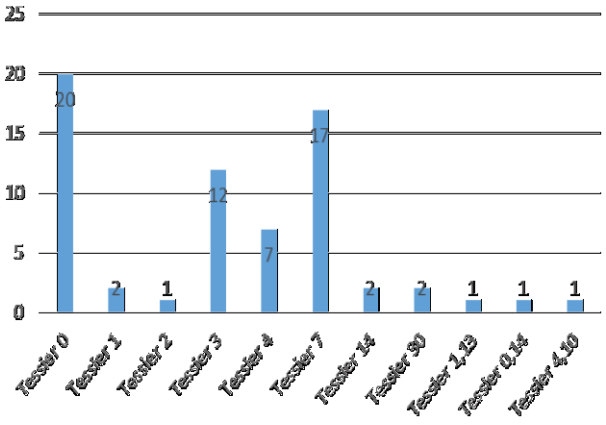

Figure 5.. Distribution of different types of Tessier clefts

In **Table 6** Tessier clefts 7 are more common in male (11/17) than female (6/17), and affect both side at the same time in most of the cases (7/17).

In **Table 7**, half of craniofacial cleft patients in our series are non syndromic (33/66). Holoprosencephaly is the most encountered syndrome in syndromic craniofacial cleft patients (22/66).

Table 6. Distribution of Tessier 7 clefts by sex and laterality			
Distribution	Male	Female	Total
Right	2	3	5
Left	4	1	5
Bilateral	5	2	7
Total	11	6	17

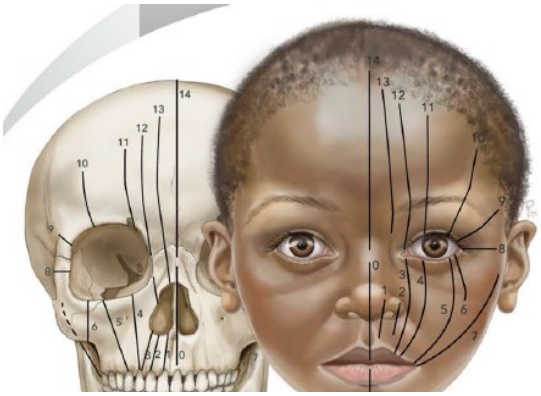

Fig 2.Tessier clefts. Image from Musolas A. et al, Cleft lip and palate-Craniofacial Clefts, 2017 Ergon (reproduced with authorisation from authors).

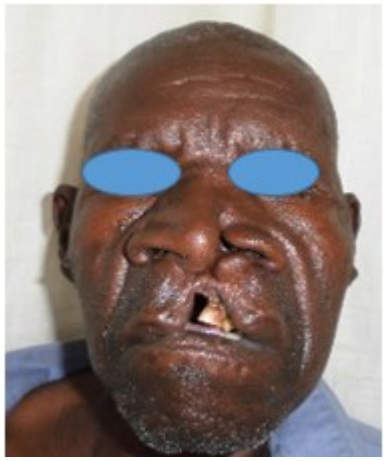

Fig. 1 An 83 years old patient with Tessier cleft 1

Fig 3. A: Tessier 0, midline anomalies and Holoprosencephaly; **B**: bilateral Tessier 7; **C**: Right side Tessier cleft 3 with left side complete cleft lip; **D**: Tessier cleft 30.

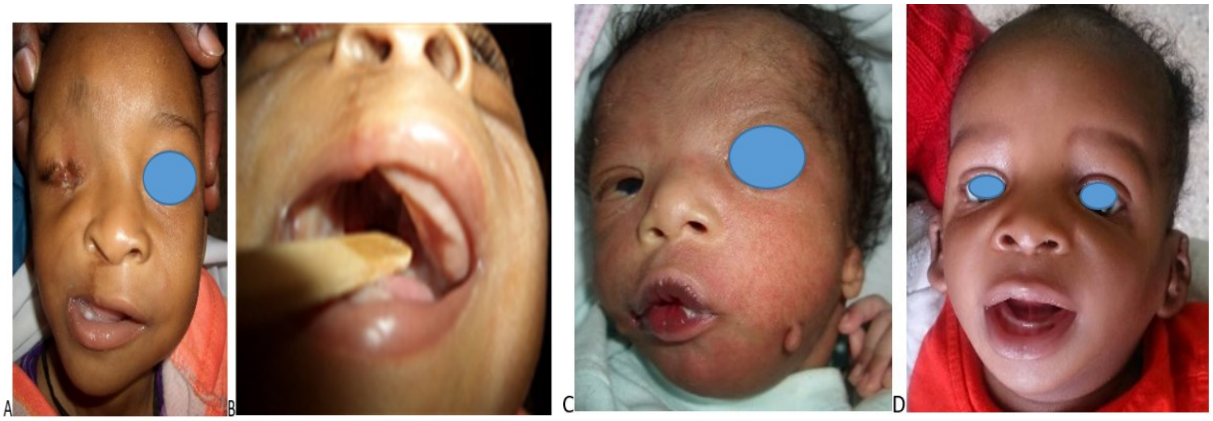

Fig 4. A, B patient with Hemifacial microsomia and Right side complete cleft lip and palate (the lip has been operated); **Fig C**: patient with Goldenhar syndrome; **Fig D**: patient with Treacher Collins Syndrome

Discussion

The aim of this study is to contribute to the general literature on rare CFC in Uganda and Africa by describing the demographic and clinical profile of CFC seen at CoRSU hospital. Most of studies on TC in the literature are limited to case studies. Those that present a series of cases are specific to a particular type of TC. So, none of them provide a general biodata and clinical profile in a big series. Our study reveals that most of children (20 cases) consult for the first time between 6 and 12 months; only 10 patients was seen before 6 months and 24 patients consult for the first time after 2 years of age (**Table1**, **Fig1**). We think probably they don't come earlier for varied reasons including lack of information, stigma, high transport costs, poor referral systems and death due to associated anomalies, malnutrition. It most likely the awareness of the condition in the society is probably not widespread.

Among orofacial clefts associated with other defects, the sex ratios shrank towards 1, 01 [1]. Our findings reveal a sex ratio of 1:1.2 with slightly more female than male but the difference is not significant (see **Table 2**). Worldwide, holoprosencephaly has female predominance [1] and 23 patients in our series are case of holoprosencephaly. Facial clefts represents the most encountered location of CFC in our series (63 cases) compared to facial clefts with extension to the cranium (3 cases) (**Fig. 2**). Probably this observation would be different if CT-Scan was done systematically for all CFC. The cost of this exam in our setting limits its use to particular cases. For more accuracy in the diagnosis of CFC, one should take in consideration the fact that the soft tissue and skeletal components are seldom affected to the same extent. The skeletal landmarks tend to be more constant and reliable than the soft tissue landmarks [2, 8]. Unilateral CFC are more common in our series (31 cases) than midline (25 cases) and bilateral (10 cases); right side (18 cases) is more common than left side (13 cases) (see **Table 4**). We didn't find in the literature any study discussing the common side of CFC in general. They are limited to specific type of TC. There are conflicting data in the literature regarding the most common type of TC: The number 3 cleft or the oro-naso-ocular cleft is presented as the most common of the Tessier CFC and number 7 cleft the most common CFC [2, 7]. Anantanarayanan P et al. reports that Tessier clefts 7 are the most common orofacial clefts, second to the more common isolated cleft lip [9]. However, large observation studies of CFC revealed that midline TC 0 is the most common of the CFC and that the combination of 0 and 14 is the most common single combination of non-isolated clefts [4, 5]. There is paucity of articles related to TC 0, and those that exist are confined to clinical examination and surgical treatment of a small number of patients [4]. Freistas R.S. et al have registered 32 cases of Tessier cleft 0 over a period of 10 years with holoprosence-phaly identified in 9 patients [4]. In our study we have registered 25 cases of midline clefts of which 20 cases were TC 0, two cases of Tessier 14 and one case of Tessier 0,14 (all associated with holoprosence-phaly) and two cases of Tessier 30 (**Fig. 5**, **Fig. 3**). As noted in this study, we too believe that TC 0 are the most common type of TC; however, with respect to the laterality of the cleft, unilateral Tessier clefts are more common than midline and right side more diagnosed. Transverse facial cleft (Tessier cleft 7) or congenital macrostomia is a rare congenital anomaly seldom occurring alone and is frequently associated with deformities of the structures developing from the first and second branchial arches [10]. Transverse cleft are known to be more common in males and have left predilection [10]. Makhija reported 17 TC 7 over a period of 5 years; Gokrem S. et al. reports 5 patients over a

period of 17 years [11]. In this context, our report of 17 cases over a period of 11 years and 5 months shows that TC 7 are common in male and have a bilateral predilection (**Table 6**, **Fig. 3B**). TC 7 is seen in some cases of craniofacial microsomia (oculo-auriculo-vertebral spectrum).It is also seen in Treacher-Collins syndrome [2]. In our series, out of the 17 TC7, four were associated with hemifacial microsomia while the rest were isolated TC 7. Makhija et al. reported a series in which 7 out of 17 were non-syndromic and isolated transverse clefts [10]. In most of the reported series, majority of the TC 7 are associated with hemifacial microsomia [10]. Hemifacial microsomia is the second most common developmental craniofacial anomaly after cleft lip and palate. The incidence of the hemifacial microsomia is between 1 in 3000 and 1 in 5600 births. Males are more frequently affected than females and the right side is affected more often than the left [12, 13]. Goldenhar syndrome is a variant of HFM, which includes vertebral anomalies and epibulbar dermoids. This is known as craniofacial microsomia when there is involvement of cranial deformities [13]. In general in our series 50% of CFC are non syndromic and 50% syndromic associated with either holoprosencephaly (23 cases), hemifacial microsomia (6 cases), Goldenhar syndrome (3 cases) or Treacher collins syndrome (2 cases) (**Table 7**, **Fig. 4**). TC 4 is a rare, complex, and challenging craniofacial malformation [14]. In our series we have registered 7 cases of TC 4 in 11 years and five months (**Figure 5**) whereas Alonso N et al. have reported 21 cases of Facial clefts 4 in 20 years. Literature review has revealed few patients with number 4 facial cleft, which reflects the rarity of this malformation [14]. TC 1-13 is an atypical CKC with with an choanal atresia [15]. We have registered only one case of Tessier 1 through 13 without choanal atresia (Table 3). Bonafos G. et al emphasize the interest of looking for evidence of a choanal atresia in any case of cleft number 13 through 1 or 12 through 2. This association is empirically justified by clinical study and reinforced by the embryopathoge-nic data [16].

No cases of Tessier 5, 6 and isolated cranial cleft from 8 to 13 were encountered in our data. Median cleft of mandible and lower lip (TC 30) is rare. We registered 2 cases over a period of 11 years and 5 months (**Figure 5**, **Fig 3D**). Only a few cases have been reported in the literature with different variations till now [17].

Table 7. Distribution of syndromes associated with Tessier clefts

Associated syndrome	Number
TCS	2
HFM	6
GS	3
HPE	22
No syndrome	33
Total	**66**

Acronyms: TCS: Treacher Collins Syndrome, HFM: Hemifacial Microsomia, GS: Goldenhar Syndrome, and HPE:

Conclusion

CFC are a rare set of malformations for which there is a paucity of literature that is often limited to isolated case reports with little objective long-term follow up [5]. Most patient in our setting consult late; we have registered slightly more female than male with a sex ratio of 1:1.2. There are conflicting data in the literature regarding the most common type of TC: midline TC 0 is the commonest clinical type of CFC seen in our setting. TC 7 is common in male and has a bilateral predilection. Fifty percent of CFC in our

series is syndromic. There is a big need to conduct a study with a larger series including CT-Scan in order to analyze more accurately clinical findings.

Contribution details

Tshimbila K. [COSECSA FCS trainee in Plastic Surgery - General Surgeon , Department of Surgery - University of Goma/DRCongo]: Conception, design, definition of intellectual content, literature search, data acquisition, data analysis, statistical analysis, manuscript preparation, manuscript editing and manuscript review.

Hodges A. M. [Consultant in Plastic surgery in charge of training at CoRSU hospital – Mbarara University/Uganda]: Definition of intellectual content, data acquisition, manuscript editing and manuscript review

Galiwango G.W.[Consultant in Plastic Surgery and Head of Department of Plastic Surgery at CoRSU hospital – Mbarara University/Uganda]: Definition of intellectual content, manuscript editing and manuscript review.

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