IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 22, Issue 8 Ser.3 (August. 2023), PP 20-24
www.iosrjournals.org

Management Of Patients With Congenital Anomolies Of Oro – Facial Deformities In Zliten, Libya

Musbah Ashmmam
Department Of Maxillo-Facial Surgery, Zliten Teaching Hospital, Zliten, Libya.

ABSTRACT:
Orofacial clefts are among the most common congenital craniofacial malformations and may be associated with other birth defects. However, the proportion and type of additional anomalies vary greatly between studies. Orofacial clefts are among the most common congenital malformations of the craniofacial region, which include cleft palate only (CP) and cleft lip with or without palate (CL/P). Congenital anomalies, defined as abnormalities of structure, function, or metabolism that are present at birth, are a major public health concern due to their life threatening nature or the potential to result in disability or death. This study assessed the prevalence and type of associated congenital malformations in children with orofacial clefts, who attended the largest cleft lip and palate tertiary referral center in Portugal. The present study was investigated in Zliten Teaching Hospital, Zliten, Libya from June 2020- August 2021. Regarding gender, the group without associated malformations had 14 males 36.84% and 24 females 63.15%. whereas the group with associated malformations chromosomal syndromes with 15.78%, monogenic syndromes with 13.15%, sequence with 44.73% and MCQ of unknown causes with 26.31%. Cleft type is divided into cleft lip with 10.52% in male and 18.42% in female, cleft palate with 13.15% male and 21.05% in female, cleft lip and palate with 7.89% male and 7.89% female and others 5.26% male and 15.78% in female. This study involves the review of the various literatures on orofacial clefts, discussing the problems on the genetic basis, associated syndromes, and their management. Counselling of prospective mothers should be promoted to ensure that the abnormality is prevented at the early stages.

Keywords: *Orofacial, Cleft lip, Cleft palate, Premaxilla, Libya.*

Date of Submission: 29-07-2023 Date of Acceptance: 09-08-2023

I. INTRODUCTION:

Orofacial clefts (OFCs) are common congenital malformations of the lip, palate, or both caused by complex genetic and environmental factors. OFC may involve the lip, the roof of the mouth (hard palate), or the soft tissue in the back of the mouth (soft palate) [(1)]. OFC also involves structures around the oral cavity which can extend onto the facial structures resulting in oral, facial, and craniofacial deformity [(2)]. A cleft lip/palate may impact negatively on an individual's self-esteem, social skills, and behaviour especially among girls. Generally, boys are affected more than girls with a ratio of about 3 : 2. Males are more likely than females to have a cleft lip with or without cleft palate [(3)], while females are at a slightly greater risk for cleft palate alone. Since facial mesenchyme is derived from neural crest, it is postulated that periconceptional folic acid supplementation may reduce the occurrence of offspring with orofacial clefts [(4)]. Zinc also is important in fetal development, and deficiency of this nutrient causes isolated cleft palate and other malformations in animals; other nutrients such as riboflavin and vitamin A are also essential. Preventive efforts might entail manipulation of maternal lifestyle [(5)], improved diet and use of multivitamin and mineral supplements, avoidance of certain drugs and medicines, and general awareness of social, occupational, and residential risk factors.

Although orofacial clefts most commonly appear as isolated conditions, with a generally favorable outcome for the patients, it has long been known that they may be frequently associated with other congenital malformations [(6)]. In these cases, the outcome depends primarily on the presence and type of associated malformations [(7)]. However, the proportions of patients with orofacial clefts with additional abnormalities varies greatly between studies, from 1.5% to 64.2%. Also, there is no consensus on the type of malformations that are most commonly associated with orofacial clefts [(8)]. The interplay of different environmental and genetic risk factors has been proposed as an underlying mechanism for orofacial clefts. However, a single major risk factor for these congenital malformations has not been identified yet, suggesting a more complex etiology than the oligogenic model originally proposed [(9)]. Moreover, consanguinity and a positive family history for orofacial clefts also play a role. Those whose parents have a close degree consanguinity and those with a positive family history for clefts are subject to higher risks for congenital malformations [(10)]. Hence, the identification of specific co-occurring congenital malformations with orofacial clefts is important for improving the definition of the etiology of this pathology.

Treatment of cleft lip and palate anomalies requires years of specialized care and is costly. The average lifetime medical cost for treatment of one individual affected with a cleft lip and palate is $100,000. Although successful treatment of the cosmetic and functional aspects of orofacial cleft anomalies is now possible, it is still challenging, lengthy, costly, and dependent on the skills and experience of a medical team. This especially applies to surgical, dental, and speech therapies [(11)]. The mean and median costs for children ≤10 years of age with an orofacial cleft were eight times higher than those for children of the same age without an orofacial cleft [(12)]. Mean costs for infants with a cleft and another major unrelated defect were 25 times higher than those for an infant without a cleft and five times higher than those for infants with an isolated cleft with patients continuously enrolled in a fee-for-service.

Services and treatment for children with OFCs vary depending on the severity of the cleft; the presence of associated syndromes, other birth defects, or both; and the child's age and needs. Orofacial clefts generally require surgical repair [(13)]. Often multiple surgeries are needed to reconstruct the lip and palate. A palatoplasty is the procedure utilized to close the palate, restore the velopharyngeal sphincter, and help speech function and other processes. The optimum approach to the treatment of children born with cleft defects is a multidisciplinary approach which involves combined efforts of a pediatrician, orthodontist, specialist nurse, cleft surgeon, speech therapist, and ear, nose, and throat specialist to provide the best combined expertise to ensure that the correct interventions are carried out at the appropriate time and to ensure the best functional and aesthetic result [(14)]. Many children will need additional surgeries as they get older. Surgical repair can improve the look and appearance of a child's face; it also may improve breathing, and shearing, speech and language. The psychological care of the patient with a cleft begins at the time of diagnosis, even if this is before birth [(15)]. An accurate diagnosis is critical to the process of counseling families. It is the responsibility of the referral centre to define the nature of the structural defect with as much precision as possible. This helps the family to visualize the child and to discuss feeding, especially breastfeeding. It also helps when informing about timing and type of surgery. To plan for the future, parents need to discuss the management and likely the treatment pathway at their own pace and at their own time, so that they are able to absorb the information. Delayed repair of cleft can lead to impaired family and societal relationships with potential long-term psychological effects on the child [(16)]. As the child matures and faces the task of individuation from the family, there may be a need for psychological work, and since adulthood provides its own set of challenges to the individual, there is potential for further psychological interventions throughout this period of life [(17)]. Parents need reassurance, support, and time to assimilate the information to be able to provide the child with the support and care needed.

METHODOLOGY:

Study Place:

This retrospective study was conducted in Department Of Maxillo-Facial Surgery, Zliten Teaching Hospital, Zliten, Libya.

Study period:

The study period conducted from June 2020- August 2021.

Sampling Procedure:

Data were collected from the Cleft Patient Data Sheet, usually completed by the physician in the first appointment by direct interview of the patient or parents and by physical examination. Data were also collected from all available patients' medical records (electronic and paper), including prenatal consultation, maternity, neonatal unit, outpatient clinic, pediatrics, and pediatric surgery files. Variables under study included the following: date of birth, sex, follow-up period, occurrence and laterality of the orofacial cleft, associated malformations and respective molecular diagnosis, family history of orofacial clefts, consanguinity between the parents, and prenatal ultrasound diagnosis.

Orofacial clefts were described according to Tessier's anatomical classification. Their occurrence was categorized as unilateral or bilateral, and complete, incomplete, or microform (eg, submucous cleft palate). Cases of orofacial clefts were categorized as: without associated malformations, whenever no other congenital abnormalities were identified; or with associated malformations, whether 1 or more congenital abnormalities, unrelated to orofacial clefts, were also present. Dental anomalies were excluded from this study as associated malformations because most of these anomalies are closely related to orofacial clefts. Cases of orofacial clefts with associated malformations were further divided into 4 categories according to their etiology: recognized causes, such as chromosomal syndromes (ie, involving clinically significant structural and/or numerical chromosomal abnormalities), monogenic syndromes (ie, related to a single gene), or sequence (ie, occurrence of associated anomalies due to a single known structural defect), or multiple congenital anomalies (MCAs) of unknown origin. For this study, MCA cases were defined as cases with 2 or more structural malformations (other

than the cleft) that could not be explained by an underlying syndrome or sequence. The MCA were grouped according to the organ system or the anatomic region primarily affected.

Statistical Analysis:

The collected data were analyzed using the SPSS software (version 20.0). Continuous variables were summarized by mean and minimum-maximum. Categorical variables were expressed as number and percentage of cases in each group (ie, with and without associated malformations) and compared using the Chi-square test or Fisher's exact test, as appropriate. Due to; the study design, no sample calculation was performed.

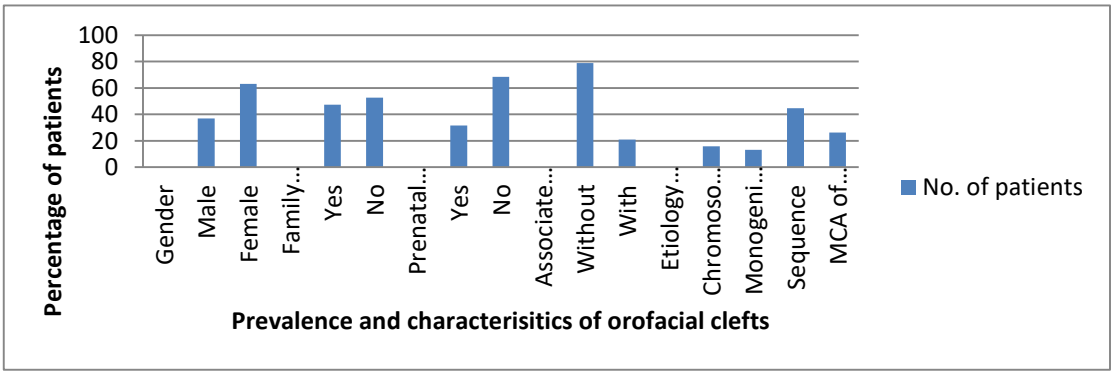
II. RESULT AND DISCUSSION:

Orofacial cleft is one of the commonest congenital irregularities which impacts adcersely on the life of the individual and to an enormous influences the family. Brought about by the collaboration of ecological and hereditary, this variation from the norm realizes diminished personal satisfaction. Only the data 34 patients were included in our analysis. Of those patients, 14 (36.84%) were males and 24 (63.15%) were females. Patients were followed up until a mean age of 15 years old (minimum 1 year and 2 months untill maximum 33 years).

Table 1: Prevalence and characteristics of Orofacial Clefts in the study population.

Characteristics	No. of patients (n=38)	Mean	P-Value
Gender			
Male	14	36.84	
Female	24	63.15	
Family history of Clefting			
Yes	18	47.36	
No	20	52.63	
Prenatal ultrasound diagnosis			0.0912
Yes	12	31.57	
No	26	68.42	
Associated malformations			
Without	30	78.94	
With	08	21.05	
Etiology of associated malformation			
Chromosomal syndrome	06	15.78	
Monogenic syndrome	05	13.15	
Sequence	17	44.73	
MCA of unknown causes	10	26.31	

(% calculated from 38 patients)



Graph 1: Prevalence and characteristics of Orofacial Clefts in the study population

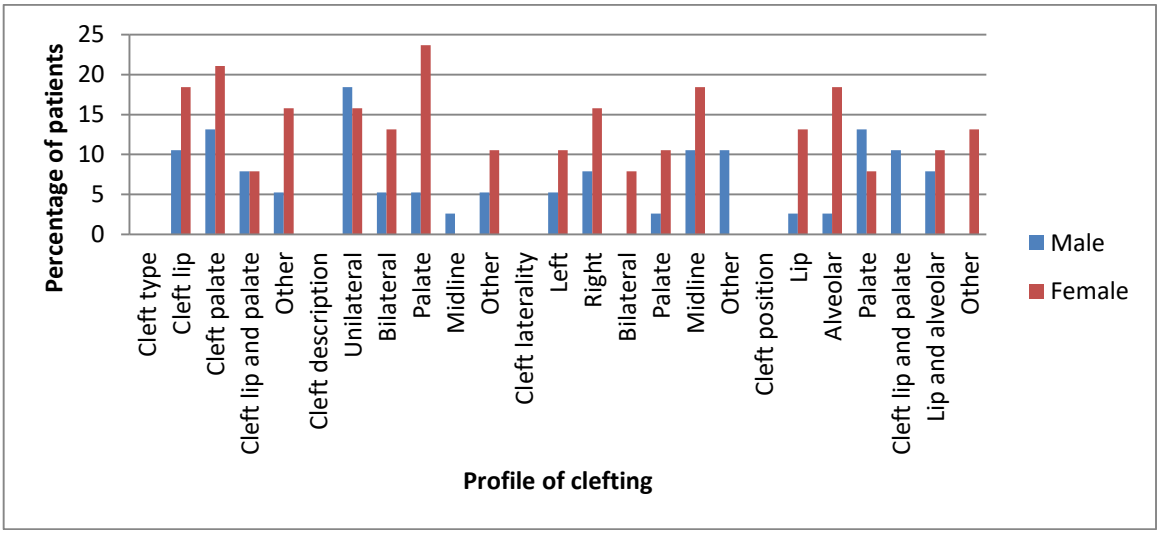
The prevalence and characteristics of the orofacial clefts and associated malformations are shown in Tables 1. Regarding gender, the group without associated malformations had 14 males (36.84%) and 24 females (63.15%), whereas the group with associated malformations chromosomal syndromes with 15.78%, monogenic syndromes with 13.15%, sequence with 44.73% and MCQ of unknown causes with 26.31%.

Table 2: Profile of clefting at patients in hospital.

Description	Gender					
	Male		Female		Total	
	No.	%	No.	%	No.	%
Cleft type						
Cleft lip	04	10.52	07	18.42	11	28.94
Cleft palate	05	13.15	08	21.05	13	34.21
Cleft lip and palate	03	7.89	03	7.89	06	15.78
Other	02	5.26	06	15.78	08	21.05
Cleft description						
Unilateral	07	18.42	06	15.78	13	34.21
Bilateral	02	5.26	05	13.15	07	18.42
Palate	02	5.26	09	23.68	11	28.94
Midline	01	2.63	00	00	01	2.63
Other	02	5.26	04	10.52	06	15.78
Cleft laterality						
Left	02	5.26	04	10.52	06	15.78
Right	03	7.89	06	15.78	09	23.68
Bilateral	00	00	03	7.89	03	7.89
Palate	01	2.63	04	10.52	05	13.15
Midline	04	10.52	07	18.42	11	28.94
Other	04	10.52	00	00	05	13.15
Cleft position						
Lip	01	2.63	05	13.15	06	15.78
Alveolar	01	2.63	07	18.42	08	21.05
Palate	05	13.15	03	7.89	08	21.05
Cleft lip and palate	04	10.52	00	00	04	10.52
Lip and alveolar	03	7.89	04	10.52	07	18.42
Other	00	00	05	13.15	05	13.15

(% calculated from 38 patients)

Table 2 is tabulated with Profile of clefting, in the table cleft type is divided into cleft lip with 10.52% in male and 18.42% in female, cleft palate with 13.15% male and 21.05% in female, cleft lip and palate with 7.89% male and 7.89% female and others 5.26% male and 15.78% in female. In cleft position is classified as lip, alveolar, palate, cleft lip and palate, lip and alveolar and others with male (2.63%, 2.63%, 13.15%, 10.52% and 7.89%) and female (13.15%, 18.42%, 7.89%, 0, 10.52% and 13.15%) respectively.



Graph 2: Profile of clefting at patients in hospital.

III. CONCLUSION:

This study provides a basis for research of the etiology of orofacial clefts. The presence and nature of different synchronous malformations might indicate different mechanisms of abnormal prenatal development. Identification of smaller subgroups or clusters may be important in etiological studies to elucidate the environmental and genetic risk factors and the interaction between them.

The overall prevalence of associated malformations (nearly 1 in 3 infants) emphasizes the need for a more comprehensive evaluation of children with orofacial clefts. An early screening routine for other congenital malformations, particularly those of the head and neck, cardiovascular, skeletal, and central nervous systems, should be considered in all orofacial clefts patients, especially when considering lip surgery within the first days of life, as many severe defects may not be diagnosed during the neonatal period by clinical examination alone. Genetic counseling might be also valuable, particularly in the orofacial cleft cases with associated malformations. Strict cooperation between cleft team members is essential to comprehensively cover all aspects of the management of the patient with orofacial clefts.

REFERENCES:

[1]. Eshete M, Butali A, Deressa W, Pagan-Rivera K, Hailu T And Abate F (2017) Descriptive Epidemiology Of Orofacial Clefts In Ethiopia. Journal Of Craniofacial Surgery, 28(2):334–337.

[2]. Mbuyi-Musanzayi S, Kayembe TJ, Kashal MK, Lukusa PT, Kalenga PM, Tshilombo FK, Devriendt K, Reychler H (2018) Non-Syndromic Cleft Lip And/Or Cleft Palate: Epidemiology And Risk Factors In Lubumbashi (DR Congo), A Case-Control Study. Journal Of Cranio-Maxillofacial Surgery, 46(7):1051–1058.

[3]. Bamford L, Mckerrow N, Barron P And Aung Y (2018) Child Mortality In South Africa: Fewer Deaths, But Better Data Are Needed. South African Medical Journal, 108(3):25–32

[4]. Hlongwa P And Rispel LC (2018) "People Look And Ask Lots Of Questions": Caregivers' Perceptions Of Healthcare Provision And Support For Children Born With Cleft Lip And Palate. BMC Public Health, 18(1):506.

[5]. Nagalo K, Ouédraogo I, Laberge J-M, Caouette-Laberge L And Turgeon J (2015) Epidemiology, Clinical Aspects And Management Of Cleft Lip And/Or Palate In Burkina Faso: A Humanitarian Pediatric Surgery-Based Study. Open Journal Of Pediatrics, 5(2):113–120.

[6]. Benjamin RH, Navarro Sanchez ML, Chen H, Mitchell LE, Langloi PH, Canfield MA, Swartz MD And Scheuerle AE (2019) Co-Occurring Defect Analysis: A Platform For Analyzing Birth Defect Co-Occurrence In Registries. Birth Defects Research, 111(18):1356–1364.

[7]. Pereira AV, Fradinho N, Carmo S, De Sousa JM, Rasteiro D, Duarte R And Leal MJ (2018) Associated Malformations In Children With Orofacial Clefts In Portugal: A 31-Year Study. Plastic And Reconstruction Surgery Global Open, 6(2):E1635.

[8]. Olusanya AA And Akadiri OA (2020) Congenital Syngnathia: Review Of Clinical Profile And Proposal Of A New Classification – Based Management Protocol. International Journal Of Oral And Maxillofacial Surgery, 49(4):505-514.

[9]. Ray S, Golden WC And Walsh J (2019) Anatomic Distribution Of The Morphologic Variation Of The Upper Lip Frenulum Among Healthy Newborns. JAMA Otolaryngology – Head And Neck Surgery, 145(10):931-938.

[10]. Hlongwa P, Levin J And Rispel LC (2019) Epidemiology And Clinical Profile Of Individuals With Cleft Lip And Palate Utilising Specialised Academic Treatment Centres In South Africa. Plos One, 14(5):1–14.

[11]. Barrett W, Diedericks B And Barrett C (2019) Congenital Heart Defects In Children With Cleft Lips And/Or Palates At An Academic Hospital In Central South Africa. South African Journal Of Anaesthesia Analgesia, 25(2):23–29.

[12]. Basha M, Demeer B And Revencu N (2018) Whole Exome Sequencing Identifies Mutations In 10% Of Patients With Familial Non-Syndromic Cleft Lip And/Or Palate In Genes Mutated In Well-Known Syndromes. Journal Of Medical Genetics, 55(3):449–458.

[13]. Zhang J, Zhao H And Huang W (2020) A Novel FZD6 Mutation Revealed The Cause Of Cleft Lip And/Or Palate In A Chinese Family. Genes And Diseases, 7(3):440-447.

[14]. Worley ML, Patel KG And Kilpatrick LA (2018) Cleft Lip And Palate. Clinical Perinatology, 45 (4):661-678.

[15]. Tian H And Feng J (2017) Intraflagellar Transport 88 (IFT88) Is Crucial For Craniofacial Development In Mice And Is A Candidate Gene For Human Cleft Lip And Palate. Human Molecular Genetics, 26 (5):860-872.