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Original Article

Insight into the Patterns and Misconceptions Associated with Cleft Lip and Palate in the Local Population

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Abstract

Objective: This study aimed to get an insight into the patterns and misconceptions associated with Cleft Lip and Palate (CL&P) in the local population.

Methods: A total of 104 children of 3 months to 20 years of age with non-syndromic cleft lip and palate, isolated cleft lip (iCL), or isolated cleft palate (iCP) were enrolled. General information such as age, gender, cast, and clinical information consisting of the type of cleft and site of involvement was taken. Parents of all patients were asked for consanguinity and affected members in family and relatives, their opinion for its occurrence, and folic acid intake by mothers.

Results: Mean age was found to be 72.3+44 months with an age range of 03 months to 13years. There were 52 males and an equal number of females. CL&P was present in 57 children and bilateral involvement was seen in 32 cases. Among 104 children, positive family history was seen in 18 children with CL&P, iCL, and iCP. Folic acid was taken by the mothers of only 8 children out of 104 affected children. The majority of parents 67(64.4%) attributed CL&P to lunar or solar eclipses, and 17(16.3%) to black magic.

Conclusion: Cleft lip and palate are more common than cleft lip or palate only and may present as bilateral or unilateral. CL&P children suffer social stigmatization, discrimination, and bullying due to their appearance, therefore it is important to raise awareness among the people in order to reduce the misconceptions.

Keywords: Cleft lip and palate, Patterns, social stigmatization

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Introduction

Non-syndromic cleft lip and palate (NSCL&P) is the commonest disorder of the craniofacial region around the globe with a variable frequency based on ethnicity and country. In Pakistan, the crude birth rate of CL&P is 25.6 per 1000 population, while in India it is 20 and about 13 in China per 1000 population.[1] Asian population have the highest incidence of cleft lip and palate (0.82 to 4.04 / live births) and Caucasians and Africans show intermediate (0.9 to 2.69/1000 live birth) and low (0.18 to 1.67 /1000 live births) incidence respectively. While it affects 1.76 and 0.85 to 2.68 per 1000 live birth in Chinese and Japanese populations respectively.[2]

Non-Syndromic and syndromic variants exist with syndromic type associated with additional distinctive features along with cleft lip or palate while NSCL&P have no other observed congenital deformity elsewhere in the body.[3] CL&P can occur as unilateral clefts involving one side or bilateral clefts affecting both sides, former is more common than bilateral clefts with a ratio of 4:1, and the left side of the face is affected 70% more than the right side. Cleft lip and palate are considered as multifactorial condition associated with many risk factors including genetics or folic acid intake by mothers during pregnancy. As literature shows the decreased risk of developing CL&P in children whose mother taking Folic acid during early phase of their pregnancies.[4]

Also, there is a variation among different countries about cultural belief of CL& P causation. In Western countries, it is being accepted CL&P may be caused by nature while in the low-income countries' persons are blamed for the occurrence of this disease because of their sins or as punishments. The child born with CL&P are not treated well leading to destructive results.[5] Therefore,

it is a need of time to understand the background of affected parents and children and provide them better health facilities globally.

The current study aimed to determine the pattern of cleft lip and palate and the misconception about its occurrence in local population. In addition to facilitating early diagnosis, it may create awareness, reduce stigmatization and enable affected individuals and their families to benefit from health care facilities.

Methods

This descriptive study was conducted in the department of Oral Pathology University of Health Sciences Lahore after taking approval from Institutional Review Board from February 2020- December 2020. A total of 104 children of 3 months to 20 years of age with non-syndromic cleft lip and palate isolated cleft lip, or isolated cleft palate from Cleft Lip and Palate (CLAP) Hospital and camps arranged by CLAP Hospital in different provinces of Pakistan with a confirmed clinical diagnosis were enrolled. However, patients with any other deformity along with cleft lip or palate were excluded. Written informed consent was obtained from the parents of each participant. General information such as age, gender, cast, and clinical information consisting of the type of cleft and site of involvement was taken on a specially designed data collection form. Parents of all children were asked for consanguinity and affected members in family and relatives, their opinion for its occurrence, and folic acid intake by mothers.

Statistical Analysis: SPSS (26.0) was used to analyse the data. Mean+ SD was determined for age. Frequency and percentages were calculated for gender, cleft lip and palate phenotype and laterality. Chi-square test was applied to observe the association between gender and cleft lip and palate phenotypes. P value equal to or less than 0.05 was considered as statistically significant.

Results

There were 52 males and an equal number of females in 104 children with NSCL&P. Mean age was found to be 72.3+44 months with an age range of 3months to 13years. CL&P was present in 57(64.8%) children and bilateral involvement was seen in 32(30.8%) children as presented in table 1.

Table 1: *Distribution of cleft lip and palate phenotypes and laterality*

Cleft lip and palate type	Frequency (n)	Percentage (%)	Total
Isolated cleft lip	27	26	104
Isolated cleft Palate	19	18.2	
Cleft lip and Palate	57	54.8	
Laterality			
Unilateral-Right	22	21.2	85*
Unilateral-left	31	29.8	
Bi-lateral	32	30.8	

* Laterality was not determined in 19 cases of Isolated cleft palate.

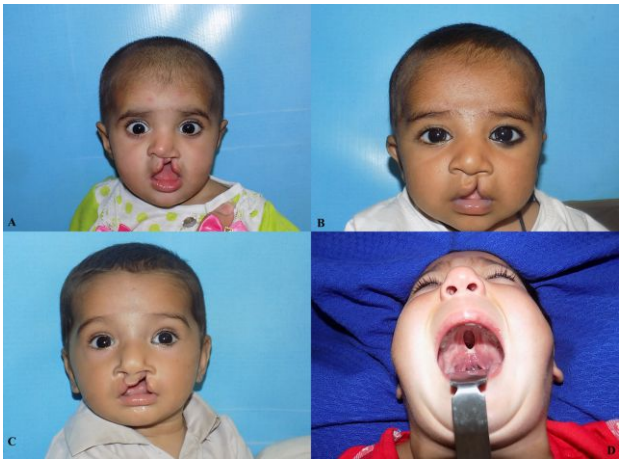

Figure 1: *This figure shows different types of cleft lip and palate. (A&B) unilateral cleft lip and palate, (C) bilateral cleft lip and palate, (D) cleft of hard palate only.*

In Asian countries, like Pakistan, cousin marriages are quite common. Parents of children were asked for cousin marriages and there were 88% of cousin marriages. Among 104 children, positive family history was seen in 18 children with CL&P, iCL, and iCP. Affected family members were first-degree relatives including brother, father, father's sister, and mother's sister.

Folic acid was taken by the mothers of only 8 children out of 104 affected children. Few of them never heard about the folic acid intake. Some of them denied the role of folic acid with CL&P altogether. The majority of parents 67(64.4%) attributed CL&P to lunar or solar eclipses, 17(16.3%) to black magic, and 20 (19.2%) didn't ascribe a cause to CL&P.

Chi-square test was applied to determine the association of demographic and clinical variables. A significant association between gender and cleft phenotype was observed. Positive family history also showed a significant association with CL&P phenotype as indicated in table 2. This significant association showed the importance of family history in determining the incidence of inheritance of CL&P in affected families.

Discussion

Cleft lip and palate are commonly occurring structural birth defects with a spectrum of phenotypes. Low mortality but high rate of morbidity is present in infants affected with NSCL&P. These children suffer with multiple challenges in their early life, including difficult to eat, breath, hear, dental problems and frequent infections.

Table 2: *Association between demographics and cleft lip and palate type and laterality*

Variables	Gender		p-value	Family History		p-value	Consanguineous		p-value	Folic acid intake by mother		p-value
	Male n (%)	Female n (%)		Positive n (%)	Negative n (%)		Yes n (%)	No n (%)		Yes n (%)	No n (%)	
Cleft lip only	12(44)	15(56)		8(30)	19(70)		23(85)	4(15)		3(11)	24(89)	
Cleft Palate only	6(30)	14(70)	**.05**	5(25)	15(75)	**.03**	16(80)	4(20)	**.76**	2(10)	18(90)	**.53**
Cleft lip and palate	34(60)	23(40)		5(9)	52(91)		49(86)	8(14)		3(5)	54(95)	
Unilateral (right)	14(64)	8(36)		2(9)	20(91)		20(91)	2(9)		0	22(100)	
Unilateral (left)	12(39)	19(61)	**.05**	6(19)	25(81)	**.52**	26(84)	5(16)	**.78**	4(13)	27(87)	**.30**
Bi-lateral	20(63)	12(37)		5(16)	27(84)		27(84)	5(16)		2(6)	30(94)	

Chi-Square was applied to determine the P-value.

Disturbances in morphogenesis of normal face result in NSCL&P. The process of development of palate is a complex process mediated by signalling molecules, cell-surface receptors, and growth factors networks.

Smile Train a non-profit organization provided the data of 25 centres of Pakistan performing 50,000 surgeries to repair cleft lip and palate from 2008 to 2015 and more than 8000 clefts were repaired in 2014 alone. There is a limited data about the prevalence of cleft lip ad palate in Pakistan except few studies conducted on smaller scale.[6] In 2013, an isolated study conducted in Karachi revealed CL& P as the most prevalent birth defect with a hesitation in looking for care and lack of follow-up after surgical repair, no involvement of speech therapists and lack of Orthodontists.[7] Likewise, another study conducted on small-scale in Peshawar showed 1.91 /1000 births. In current study, family history and folic acid intake by mother during pregnancy was also determined.

Current study participants had a mean age of 72.3 + 44 months which is an older cohort as compared to another study which reported mean age as 41 ± 54 months.[8] International studies have reported participants from birth to 40 years however highest reported cases are documented in age groups of 1 year to 10 years.[9] Reason for this variation reflects the time usually most parents seek treatment for their children.

There was equal gender distribution of the participants in the present study among all phenotypes. Slightly higher male involvement has been hinted ranging from 51% to 55% in some of the studies on the subject of CL&P. Also, a study from Pakistan reported more males (68.2%) affected than females (32%).[10]

Regarding the distribution of cleft types among genders, CL&P was seen more in males than females and cleft lip only and cleft palate only was observed more in females in present study similar to previous findings.[11] Cleft lip alone is more frequent than isolated cleft palate and combined CL& P deformities. An increased prevalence of CL&P in males and cleft palate alone in females has also been reported.[12] Another study described the gender-specific distribution of iCL and CL&P with a male to female ratio of 1.7:1.0 and 1.4:1.0 respectively[13]. This was reversed in case of iCP with females affected more than males with a ratio of 1.6:1.0.

Literature from Pakistan reports cleft lip alone to have higher frequency in males while females presented more with cleft palate only. However, study from the Northern regions of the country have reported cleft lip and palate alone to be higher in females than males again highlighting the variation in population studied and selected.[14] The reason for this variation can be because of genetic differences in both genders. Palatal shelves arise to horizontal position from vertical approximately half a week later in females than in male embryos thereby increasing the risk for iCP.[15] Gender therefore is considered as a non-modifiable risk factor of occurrence for orofacial clefts.

Regarding laterality, majority of the research found more frequent unilateral cleft than bilateral having a ratio of 4:1, and about 70% affect the left side of the face. This is validated further in the current findings. About 40% of cases are directly linked to genetic factors with a familial tendency.[4] The same findings were observed in current study. However, in contrary to present study bilateral cleft cases were found more than unilateral clefts.[10] Among unilateral clefts, left side is most commonly affected than right side with or without cleft palate, with no specific reason. However, one possible explanation can be increased blood supply of the right side of the fetal head as supplying blood vessels the leave the aortic arch closer to the heart, this results better nourished right side than left side as suggested by Johnston.[16] Variations in laterality are attributed to fluctuating and directional asymmetry. Facial directional asymmetry is also more commonly seen in relatives of children with clefts while comparing with the general population.

In regard to phenotypes, multiethnicity reflects variations in different types of clefts. Multifactorial threshold (MFT) model prophies greater genetic variations within

a population having a higher prevalence of cleft prevalence that may lead to occurrence of severe cleft types.[17] In a Singaporean study, various ethnicities reported CL & P as most common orofacial clefts among live births with a distribution of 2.2 for iCP, 2.1 for CL&P and 1.0 for iCL.[18] The Global Registry and Database on Craniofacial Anomalies reports low prevalence of iCP and high rates of CL&P with a ratio 4:6 in Asia according to the findings of Asian OFC registries.[19] Other studies from Iran and Italy found iCP more than CL&P and iCL.[20,21] It was suggested that iCL and CL&P share same etiological spectrum with different severities, but the evidence was not decisive. Both of these iCP and CL&P possess different aetiology hence classified separately.

CL&P and iCP have different developmental origins of the lip or primary palate and the secondary palate with diverse cellular and genetic aetiologies. Isolated cleft palate may arise independently or secondarily from cleft lip. Epidemiologic data suggests distinct aetiologies features of iCL from cleft lip with palate and therefore, it is classified accordingly.[22]

Isolated CL accounts 25% of all clefts, while cleft lip and palate both comprise of 45%. iCL and CL&P affect the boys more commonly whereas cleft palate only is found predominantly in females. Various studies have found health care services and an access to CL&P treatment, and interaction and strong communication among healthcare team members is required for the effective the treatment of CL&P.[23]

Positive family history increases risk of having a NSCL &P child by 3-5%.[24] Positive family history was seen in 18% of the children in present study. Studies from Arab, Asian, and European countries have observed a positive family history ranging from 20% to 28% which is slightly higher than the present findings.[20] These differences can be due to the variation in sample sizes among the studies. Consanguineous marriages are considered beneficial socially, therefore preferred by more than 20% of the world population.[25] It positively supports the women in patrilineal descent of families in the developing countries. A highest rate of consanguinity of 73% and 5%–60% has been found in Pakistan and India respectively.[26,27] Around the world different studies have reported a range of cousin marriages in affected children to from approximately 20% to 60%. Current study found almost 90% of the parents of the study participants to be maternal or paternal cousins who is highest reported from the region. There is a high genetic risk to develop congenital disorders in consanguineous couples. It is, therefore, required to develop future health care programs targeted at screening the carriers.[28]

Pre-conceptual and early pregnancy intake of folic acid by mothers is considered as a preventive measure against neural tube defects however a consensus is still not developed among researchers.[29] Meta-analyses, however, support folic acid intake as a potential preventive measure against NSCL&P.[30] Mothers of only 8 children took folic acid in present study similar to trend in other developing countries like Syria where 98% of mothers of NSCL&P children did not take folic acid during their pregnancy. Cleft lip and palate is a biological condition that can be corrected and is most common among Caucasians and Asians. A woman with a cleft lip or palate in Africa is often blamed for practicing unfaithfulness, or for being the victim of witchcraft, and eventually punished by God.[31] However, there is no truth in these statements and these are just myths based on disbelieves. Similar to this, in Pakistan women are held responsible for all of this, and clefts are thought to be caused by lunar and solar eclipses. A typical misconception revolves around the influence of envious relatives on the mother through the use of black magic.

Conclusion

Cleft lip and palate are more common than cleft lip or palate only and may present as bilateral or unilateral with a minimal difference. A family history was not prevalent, and very few mothers took Folic acid during pregnancy and were aware of its importance. People with cleft lip and palate may face social stigma, discrimination, and bullying due to their appearance or speech difficulties. Raising awareness helps reduce the misconceptions and prejudices associated with these conditions, promoting a more inclusive society.

Limitations: The study has few limitations. The sample size was small that may not be representative of the larger population. Large samples are more likely to be representative of the broader population, improving the external validity of the study and making it easier to generalize results.

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Conflict of Interest: None

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