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ORIGINAL ARTICLE

Frequency of cleft lip and cleft palate in Pakistan.

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ABSTRACT… Objective: To find out the prevalence of patients with cleft lips and/or cleft palate and their association with risk factors. **Study Design:** Descriptive Cross-sectional study. **Setting:** Department of Plastic Surgery Jinnah Post-Graduate Medical Center, Karachi. **Period:** March 2020 to October 2021. **Material & Methods:** The patients with any age group or gender, who had cleft lip and/or palate were included in the study. The demographic details, personal and family history and clinical examination of the patients were noted on a preformed proforma. The data was analyzed by using the Statistical Package for Social Sciences (SPSS) version 20. The p-value less than 0.05 was considered as significant. **Results:** The mean age and standard deviation of the study participants was 41 ± 54 months. Male gender was predominant. History of cousin marriages were positive among 92.1% of the cases and 11.1% were having positive family history with p-value ≤0.05. Majority of cases were having both deformities including cleft lip and cleft palate with frequency of 55.2% while 30.2% of the individuals were suffering from cleft lip deformity. The affected cases of cleft palate were 14.6%. Majority of the study participants were male which predominantly reported bilateral cleft lip in association with cleft palate, followed by deformity of left cleft lip and palate. Among females the cleft palate deformity was more common (10.4%) than their male counterpart but there was no significant association with gender. **Conclusion:** Cleft deformities are one of the most common congenital anomaly in which the cleft lip associated with cleft palate is more prevalent. Majority of the affected individuals are males and mostly their parents have cousin marriages. Family history is also one of the major contributing factor.

Key words: Cleft Lip, Cleft Palate, Congenital Deformity.

INTRODUCTION

Most Common congenital abnormalities of oral and maxillofacial region are cleft lip and cleft palate. These affect children worldwide with frequency of 1 per 500 to 1 per 1000 children[1] and total number of children born every year with cleft lip and cleft palate is more then 1,60,000.[2] Highest number of children born every year with cleft lip and cleft palate are in China followed by India and then Indonesia while Pakistan lies next to Indonesia having increased number of children born with cleft lip and/or cleft palate.[3] Treatment of cleft lip and cleft palate is long term and it affects the individual psychologically, socially and physically throughout the life.[4] There is failure of fusion of medial nasal, lateral nasal and maxillary process, which usually occurs at 30 to 37 days of embryonic life, resulting in primary cleft palate while secondary cleft palate occurs because of impending position of tongue at 7th day of gestation resulting in failure of the fusion of maxillary palatal shelves.[5]

Isolated cleft lip is different genetically as well as embryologically from combined cleft lip and cleft palate. Cleft lip and cleft palate affect predominantly males while isolated cleft lip is predominant in females.[6] Cleft lip associated with cleft palate accounts 2/3rd of cases while remainder 1/3rd consist of isolated cleft lip. Patients with cleft lip in association with cleft palate have usually bilaterally deformed lip. It is a multifactorial congenital abnormality in which multiple genes are mutated along with environmental factors. There is a high risk of cleft lip and/or palate among the relatives of children

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having bilateral cleft lip in association with cleft palate. However, relatives of children with isolated cleft lip have minimal risk as compared to general population. The risk also increases, if there is history of cousin marriage among parents or history of any other congenital abnormality.[7] Cleft deformities are sometimes associated with syndromes especially isolated cleft lip occurs with multiple syndromes.[8]

The aim of current study was to find out the prevalence of patients with cleft lips and/or cleft palate and their association with risk factors.

MATERIAL & METHODS

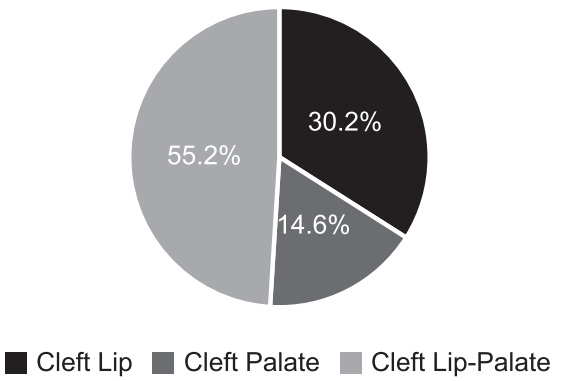
A descriptive cross-sectional study was conducted in the department of plastic surgery Jinnah Post-Graduate Medical Center, Karachi from March 2020 to October 2021. The study got approval from the institutional ethical review committee (0170617SB). The patients with any age group or gender, who had cleft lip and/or palate were included in the study. Those patients who were previously operated for the correction of cleft lip and/or palate were excluded from the study to avoid biasness in identifying the type of deformity. The demographic details, personal and family history and clinical examination of the patients were noted on a preformed proforma. Informed consent/assent was taken from the patient/guardian.

The data was analyzed by using the Statistical Package for Social Sciences (SPSS) version 20. Mean with standard deviation was calculated for numerical variables while frequency and percentages for categorical variables. To find out the association between demographic variables and cleft lip and/or palate and its types, the Chi-square was used. The p-value less than 0.05 was considered as significant.

RESULTS

About 280 patients with cleft lip/palate were studied. The mean age and standard deviation of the study participants was 41 ± 54 months, ranging from 1 month to 45 years. Male gender was predominant with frequency of 60% and were from rural area i.e. 72.9%. The two most important factors were noted with a very strong positive association including history of cousin marriages, which were positive among 92.1% of the cases and the other one is positive family history among 11.1% with p-value of 0.000 and 0.001 respectively. About 9.6% of the cases were also having history of other congenital anomalies with significant association (p-value ≤0.05). The demographic characteristics of study participants and their association with the occurrence of cleft deformities are mentioned in Table-I.

The results reported that majority of cases were having both deformities including cleft lip and cleft palate with frequency of 84 out of 280 (55.2%) while 30.2% of the individuals were suffering from cleft lip deformity. Cleft palate usually involved either the hard palate or the soft palate but can't be differentiated to right or left side. The affected cases of cleft palate were 41 (14.6%). The frequency of different types of cleft are mentioned in Figure-1.



Majority of the study participants were male which predominantly reported bilateral cleft lip in association with cleft palate, followed by deformity of left cleft lip and palate with frequency of 14.6% and 12.5% respectively as mentioned in Table-II. Among females the cleft palate deformity was more common (10.4%) than their male counterpart. Cleft lip and palate deformities were having no association with the gender as the p-values were non-significant (p-value ≥0.05).

Variables	n= 280	%	P-Value
Gender Male Female	 168 112	 60% 40%	0.61
Area Urban Rural	 76 204	 27.1% 72.9%	0.84
Cousin marriage Yes No	 258 22	 92.1% 7.9%	0.000
Family history Yes No	 31 249	 11.1% 88.9%	0.001
History of other congenital anomalies Yes No	 27 253	 9.6% 90.4%	0.039

Table-I. Demographic characteristics of study participants and their association with Cleft lip/palate

Types of Clefts		Gender		Total	Percentage	P-Value
		Male	Female			
Cleft lip	Right	21	08	29	10.4	0.52
	Left	30	17	47	16.8	
	Bilateral	03	04	7	2.6	
	Median	01	0	01	0.4	
Cleft palate		12	29	41	14.6	0.12
Cleft lip with cleft palate	Right	25	13	38	13.5	0.97
	Left	35	27	62	22.1	
	Bilateral	41	14	55	19.6	
Total		168	112	280	100	

Table-II. Frequency distribution of different types of clefts and their association with gender

DISCUSSION

Current study was conducted on about 280 cases of cleft lip and/or palate with mean age of 41 ± 54 months, consisting age range of 1 month to 45 years. Other studies from Pakistan reported variation in the age groups like Anwar M et.al. conducted study in southern Punjab on the age group of 3 months to 45 years with mean age of 39 ± 62 months[9] while Khan M et.al. took cases from Peshawar, having mean age of 3.5+6.59 years.[10] Current study reported male predominance which is consistent with other studies like one of the study conducted in Saudi Arabia by Aljohar et.al.[11] or another study done in Iran by Kianifar et.al. reported the same results of high male to female ratio.[12]

Current results reported history of cousin marriages among parents of 92.1% patients of cleft lip and/or palate deformity and a strong significant association was found. A study conducted in Southern Punjab reported higher frequency of consanguineous marriage among patient's parents that is 95.8%[9] while in Peshawar it is 61.6%$_{10+--+}$ and in Karachi Elahi MM et.al. noted frequency of cleft lip and/or palate in cousin marriages is 32%[2] and Jajja MRN et al found it in 17.1% of the cases.[13] Current study was also performed in Karachi but the variation in the results might be due to the urban population who are well-educated to understand the genetic consequences behind cousin marriages. As the study was conducted in a largest referral hospital of Sindh province so the current data

had higher percentage of cases from the rural area of Sindh where the literacy rate is very low with poor customs. Another important factor noted in the current study, was positive family history among 11.1% cases of cleft lip and/or palate with a significant positive association. The study conducted in southern Punjab found 16.6% cases with positive family history[9] while study from Peshawar found 21.4% cases.[10] Current data showed that about 9.6% of the patients with cleft lip and/or palate were having other congenital anomalies as well and this finding is also supported by the literature with variable frequencies like in Pakistan, Anwar M et.al found 12.5% cases[9], Khan M et.al. observed 17% cases of cleft lip and/or palate associated with other congenital anomalies.[10] In India Kumar et.al. noted 13.9% cases with other anomalies.[14] In Ethopia 11% of the cases were positively associated with congenital anomalies.[6] A study conducted in Iran by Kianifar et.al. reported a very high frequency of cases associated with other congenital anomalies that is 37%.[12]

Looking over the types of cleft, the current study manifested cleft lip associated with cleft palate was the most common deformity in which bilateral deformity was the most common followed by the left side involvement. These results are supported by multiple studies conducted by Anwar M et.al[9], Kianifar et.al.[12], Sharif et.al[15] and Bekele KK et.al.[6] Current study found that cleft lip with cleft palate is more common among male cases while isolated cleft palate was predominant among female cases. This finding is favored by many local studies, conducted by Anwar M et.al[9] Khan M et.al.[10] and Elahi MM et.al.[2] looking over the international studies, Kianifar et.al.[12], Prabakaran S et.al.[16], Bekele KK et.al.[6] and Kandasamy R et.al.[17] also reported higher frequency of cleft lip in association with cleft palate among male patients while isolated cleft palate deformity among female patients.

CONCLUSION

Cleft deformities are one of the most common congenital anomaly in which the cleft lip associated with cleft palate is more prevalent. Majority of the affected individuals are males and mostly their parents have cousin marriages. Family history is also one of the major contributing factor.

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2	Mohammad Farooq Bhutta	Literature review.	
3	Fatima Pervaiz Hashmi	Write first draft of manuscript.	
4	Kiran Khan	Developed period, Statistical analysis.	
5	Uzma Anam Iqbal	Manuscript writing.	
6	Zahid Iqbal Bhatti	Program design & Literature search.