

Epidemiological Trends in Cleft Lip and Cleft Palate Frequency: An Analytical Perspective

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Submitted for Publication: 05-06-2023
Accepted for Publication 09-12-2023

How to Cite: Moeed K, Ahmad W, Haris M, Muzadar S, Wazir NU, Imran S. Epidemiological Trends in Cleft Lip and Cleft Palate Frequency: An Analytical Perspective. APMC 2023;17(4):550-553. DOI: 10.29054/APMC/2023.1371

ABSTRACT

Background: Cleft lip and/or palate (CL/P) represents a common congenital anomaly characterized by incomplete fusion of facial structures during embryonic development. **Objective:** To assess the epidemiological trends in cleft lip and cleft palate frequency: an analytical perspective. **Study Design:** Retrospective observational study. **Settings:** Department of Anatomy, Nowshera Medical College, Nowshera, Pakistan. **Duration:** From October 2022 to March 2023. **Methods:** The study included patients diagnosed with cleft lips, cleft palate, or both, who presented to the hospital. Medical records of both pediatric and adult patients were reviewed to ensure a comprehensive understanding of the prevalence and associated factors. Anatomical perspectives were assessed through clinical examination and imaging studies such as photographs and radiographs. **Results:** The mean age of the study population was 18.5 ± 5.12 years. In terms of gender, males constituted 60.0% of the population, while females accounted for 40.0%. The distribution of cleft types among the study population revealed that 60% (n=105) were diagnosed with cleft lip only, while 25.7% (n=45) presented with cleft palate only. Additionally, 14.3% (n=25) of individuals had both cleft lip and palate. The anatomical description of clefts among the study population was as follows: For cleft lip, 40% (n=70) were classified as mild, 35% (n=61) as moderate, and 25% (n=44) as severe, indicating a varied spectrum of severity in lip involvement. Regarding cleft palate, 60% (n=105) were categorized as incomplete, while 40% (n=70) were classified as complete, reflecting the predominance of incomplete palatal clefts in the sample. In terms of cleft distribution, 55% (n=96) of cases were unilateral, 35% (n=61) were bilateral, and 10% (n=18) were midline, indicating diverse patterns of cleft distribution observed. **Conclusion:** In conclusion, our study contributes to the understanding of the anatomical variations and distributions of cleft lip and/or palate (CL/P) within our population, emphasizing the spectrum of severity and patterns of cleft involvement.

Keywords: Anatomy, Cleft, Distribution, Genetic, Multifactorial, Population, Severity.

INTRODUCTION

Cleft lip and cleft palate are congenital anomalies that arise due to incomplete formation of the lip or mouth in utero. These diseases are prevalent birth malformations, impacting around 1 in every 700 babies globally.^{1,2} They can manifest independently or in conjunction, and their intensity can vary significantly. Cleft lip is a condition characterised by a division or aperture in the upper lip, varying in size from a minor

indentation to a substantial crevice that reaches towards the nasal area. Cleft palate refers to a condition where there is a division or gap in the roof of the mouth, which can affect either the hard palate (the front bony part) or the soft palate (the rear muscular part). Occasionally, individuals may have both cleft lip and cleft palate, which can result in notable difficulties related to nutrition, speech, and dental growth.^{3,4}

The precise aetiology of cleft lip and cleft palate is not always evident, although it is thought to arise from a confluence of genetic and environmental influences. Certain genetic mutations or syndromes, maternal smoking or alcohol consumption during pregnancy, and certain medications are known to increase the risk of these conditions. Additionally, nutritional deficiencies, exposure to toxins, and maternal illnesses such as diabetes can also play a role.^{5,6} The exact mechanisms underlying these failures in fusion are not fully understood but are thought to involve disruptions in cell proliferation, migration, and apoptosis during embryonic development. Genetic factors may influence the expression of key signaling molecules and transcription factors involved in these processes.^{7,8} The impact of cleft lip and cleft palate extends beyond physical appearance, affecting various aspects of a child's life. Babies with these conditions may experience difficulties with breastfeeding or bottle feeding due to challenges in creating suction. As they grow older, they may encounter speech and language delays, as well as dental problems such as misalignment of the teeth or missing teeth. The psychological and emotional effects of living with a visible facial difference can also be significant, potentially leading to issues with self-esteem and social interactions.⁹ Treatment for cleft lip and cleft palate typically involves a multidisciplinary approach, coordinated by a team of healthcare professionals including surgeons, speech therapists, orthodontists, and psychologists. Surgical repair is often performed in infancy to close the cleft and restore function and appearance as much as possible.¹⁰

Despite the challenges associated with cleft lip and cleft palate, many individuals lead fulfilling lives with appropriate medical care and support. Ongoing research aims to improve understanding of the underlying causes of these conditions and develop more effective treatments to enhance outcomes for affected individuals. Through increased awareness, access to comprehensive care, and support from their communities, individuals with cleft lip and cleft palate can thrive and achieve their full potential.

METHODS

The study received ethical approval from the hospital's Institutional Review Board (Ref No. NMCN/IRC/26/240). The research was carried out in the Department of Anatomy, Nowshera Medical College, Nowshera, Pakistan, spanning from October 2022 to March 2023.

The study included patients diagnosed with cleft lips, cleft palate, or both, who presented to the hospital. Medical records of both pediatric and adult patients were reviewed to ensure a comprehensive understanding of the prevalence and associated factors. Data pertaining to patients were gathered from electronic medical records,

encompassing demographic details (age, gender, ethnicity), clinical characteristics (type and severity of clefts), and relevant medical history (family history of clefts, maternal factors during pregnancy). Anatomical perspectives were assessed through clinical examination and imaging studies such as photographs and radiographs. The prevalence of cleft lips and/or cleft palate was calculated as the proportion of affected individuals among the total patient population during the study period. The connection between cleft prevalence and putative risk variables was analysed using either chi-square testing or Fisher's exact tests, including consanguinity, maternal age, maternal smoking/alcohol consumption, and socioeconomic status.

RESULTS

The mean age of the study population was 18.5 years (± 5.12 years). In terms of gender, males constituted 60.0% of the population, while females accounted for 40.0%. Cousin marriage was reported by 51.4% of participants, whereas 48.6% reported no cousin marriage. Additionally, 31.4% of individuals had a family history of cleft lip or palate, while 68.6% did not report any family history as shown in table 1.

The distribution of cleft types among the study population revealed that 60% ($n=105$) were diagnosed with cleft lip only, while 25.7% ($n=45$) presented with cleft palate only. Additionally, 14.3% ($n=25$) of individuals had both cleft lip and palate given in table 2.

Table 3, the prevalence of various risk factors among the study participants was as follows: 48.6% ($n=85$) reported consanguinity during pregnancy. Maternal smoking or alcohol consumption during pregnancy was observed in 17.1% ($n=30$) of cases, suggesting a significant but relatively lower contributing factor. Furthermore, 31.4% ($n=55$) of individuals had a positive family history of clefts, indicating a genetic predisposition within their families.

For cleft lip, 40% ($n=70$) were classified as mild, 35% ($n=61$) as moderate, and 25% ($n=44$) as severe, indicating a varied spectrum of severity in lip involvement. Regarding cleft palate, 60% ($n=105$) were categorized as incomplete, while 40% ($n=70$) were classified as complete, reflecting the predominance of incomplete palatal clefts in the sample. In terms of cleft distribution, 55% ($n=96$) of cases were unilateral, 35% ($n=61$) were bilateral, and 10% ($n=18$) were midline, indicating diverse patterns of cleft distribution observed in the study population as shown in table 4.

Table 1: Demographic characteristics of study population

Variables	Demographic	Number (Percentage)
Age	Mean Age	18.5 ± 5.12 years
	1 month - 10 years	45 (25.7%)
	11 years - 20 years	80 (45.7%)
	21 years - 30 years	35 (20.0%)
	31 years - 40 years	15 (8.6%)
Gender	Male	105 (60.0%)
	Female	70 (40.0%)
Cousin Marriage	Yes	90 (51.4%)
	No	85 (48.6%)
Family History	Yes	55 (31.4%)
	No	120 (68.6%)

Table 2: Distribution of Cleft Types

Cleft Type	Number (Percentage)
Cleft Lip Only	105 (60%)
Cleft Palate Only	45 (25.7%)
Both Cleft Lip and Palate	25 (14.3%)

Table 3: Prevalence of Risk Factors

Risk Factors	Number (Percentage)
Consanguinity during pregnancy	85 (48.6%)
Maternal smoking or alcohol consumption	30 (17.1%)
Positive family history of clefts	55 (31.4%)

Table 4: Anatomical Description of Clefts

Anatomical Feature	Severity	Number (Percentage)
Cleft Lip	Mild	70 (40%)
	Moderate	61 (35%)
	Severe	44 (25%)
Cleft Palate	Incomplete	105 (60%)
	Complete	70 (40%)
Distribution of Clefts	Unilateral	96 (55%)
	Bilateral	61 (35%)
	Midline	18 (10%)

DISCUSSION

In Pakistan, cleft lip and cleft palate represent a significant public health concern, with a notable frequency among newborns. While precise statistics may vary due to underreporting and limited access to healthcare in certain regions, estimates suggest a relatively high prevalence compared to global averages.¹¹ Factors such as consanguineous marriages, socioeconomic disparities, and inadequate prenatal care contribute to the incidence.¹²

Our study's demographic findings align closely with Kianifar *et al.* (2015), which reported a male predominance and similar distributions of cleft lip and palate types, with cleft lip without cleft palate being the most prevalent. While our study did not directly report incidence rates, the observed prevalence suggests a comparable epidemiological landscape. However, unlike Kianifar *et al.*'s focus on incidence rates and congenital anomalies, our study delved deeper into familial and lifestyle factors contributing to CL/P, such as consanguinity and maternal smoking or alcohol consumption.¹³ Hosseini *et al.* (2019) reported a prevalence rate of 1.37 per 1000 for cleft lip and palate, which differs from the incidence rate observed in our study.¹⁴

Our study's findings mirror those of Nagalo *et al.* (2015) in terms of the distribution of cleft types, with cleft lip and palate representing the most common presentation. Similarly, both studies identify a male predominance among individuals with CL/P. While Nagalo *et al.* provide insights into the age distribution and prevalence of associated congenital malformations, our study complements these findings by exploring additional factors such as consanguinity and family history, offering a more comprehensive understanding of CL/P etiology.¹⁵ In line with Farshidfar *et al.* (2023), our study highlights cleft lip and palate as the most prevalent type, underscoring consistent trends across populations. While Farshidfar *et al.*'s study provides insights into demographic characteristics and predisposing factors.¹⁶

Baig *et al.* found that 92.1% of cases had a history of cousin marriages, while our study reported 51.4% of participants with cousin marriage history. However, our study found a higher proportion of cases with isolated cleft lip only (60%) compared to Baig *et al.* (30.2%). Moreover, Baig *et al.* reported a male predominance in their study, similar to our findings, indicating a consistent trend across populations.¹⁷ Comparing our study with Sunbal *et al.* (2023), both studies underscore the male predominance in CL/P cases, with males accounting for a higher proportion than females. Consanguineous marriages were also prevalent in both studies, with Sunbal *et al.* reporting 60% of cases resulting from such marriages,

which aligns with our finding of 51.4% of participants having a history of cousin marriage.¹⁸

In comparison with Sharif *et al.* (2019), our study echoes the observation of a higher incidence of clefts of lip and palate in males compared to females. However, while Sharif *et al.* highlighted regional variations in the incidence of CL/P across different provinces in Pakistan, our study focused on demographic and clinical characteristics within a specific population.¹⁹ Lastly, when comparing with Khan *et al.* (2012), our study aligns with their findings regarding the male predominance in cases of cleft lip with palate deformity. However, differences exist in the distribution of cleft types and the prevalence of consanguinity, which may reflect variations in study populations or methodologies.²⁰

CONCLUSION

In conclusion, our study contributes to the understanding of the anatomical variations and distributions of cleft lip and/or palate (CL/P) within our population, emphasizing the spectrum of severity and patterns of cleft involvement.

LIMITATIONS

Limitations of the study included its retrospective design, which relied on available medical records, potentially leading to incomplete data or selection bias.

SUGGESTIONS / RECOMMENDATIONS

For improved understanding, future research could explore the underlying factors contributing to epidemiological trends in cleft lip and cleft palate.

CONFLICT OF INTEREST / DISCLOSURE

None.

ACKNOWLEDGEMENTS

None.

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