Journal of Neonatal Surgery
ISSN(Online): 2226-0439
Vol. 14, Issue 19s (2025)
https://www.jneonatalsurg.com

OPEN ACCESS

Incidence Of Various Types Of Cleft Lip And Palate In Southern Kp, Pakistan

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Cite this paper as: Dr. Asif Aziz, Dr. Tauseef ul Hassan, Dr. Manzoor Ahmad, (2025) Incidence Of Various Types Of Cleft Lip And Palate In Southern Kp, Pakistan. *Journal of Neonatal Surgery*, 14 (19s), 463-474.

ABSTRACT
Cleft lip and palate (CLP) is among the most frequent congenital malformations in the world and its incidence and its etiological factors are notable among different regions. The objective of this study was to study the patterns, socioeconomic and risk factors of CLP occurring in Southern KPK, Pakistan, in which consanguinity and maternal education were hypothesized to be the important risk factors for CLP. This study was conducted from July 2017 to June 2024 over 7 years. The retrospective cross sectional study analyzed 223 confirmed CLP cases in two Tertiary Care Hospitals in districts of Bannu and Dera Ismail Khan. Cleft types, laterality, gender distribution, parental consanguinity, maternal education, family history, among other predisposing factors, were meticulously collected on a data base. Of these CLP, 1.83 per 1,000 live births were encountered, with a predominance of cleft lip with palate (46.6%), cleft lip (34.9%) and cleft palate (18.5%). Most affected patients were males (61.6%), and the majority of clefts were left sided (57.5%). Notably, consanguinity emerged as a significant risk factor, present in 83% of cases, alongside a positive family history in 14.4%. Furthermore, in 21.2% of the patients there were associated congenital anomalies, mainly congenital heart defects, and almost 19% of the children were presented to hospitals later than the infancy. These findings emphasize the multifaceted nature of CLP in this region, which includes both genetic predisposition, maternal education, and socio-economic inequality. The study recommends targeted public health strategies like avoiding consanguineous marriages, and increased access to prenatal care to reduce CLP risk. For healthcare policy and optimization of outcomes of affected children in Southern KPK, region specific insights are very critical.

Keywords: *Cleft Lip and Palate (CLP), Consanguineous Marriages, Maternal Education, Socioeconomic Factors, Epidemiology, Congenital Anomalies, Unilateral pattern.*

1. INTRODUCTION

One of the most common birth defects in the world is cleft lip and palate (CLP) occurring in about 1 in 700 of live births (Mossey et al., 2022). It is because of the lips and palate not fusing properly during early pregnancy, which causes both anatomical as well as physiological problems and noticeable facial differences (Sabbagh et al., 2023). In addition to the physical aspect, children with CLP need long term multidisciplinary care: surgery, speech therapy, psychological support (according to Berkowitz, Fasching, and Pignata, 2021). Although medical care has improved remarkably, CLP occurs more frequently and receive poorer care in some developing countries (Pakistan) (Butali & Mossey, 2022).

The statistically documented fact that prevalence of CLP is greater in Pakistan than in most Western countries, is multifactorial. It is affected by genetic background, cultural traditions for consanguineous marriage and environmental exposures (Mirza et al., 2023). Moreover, consanguinity in particular is a well-known risk factor in the region, as marrying within family increases the chance of inheriting recessive genetic mutations (Shah et al., 2022). Furthermore, in addition to all of that above, congenital malformations are stimulated by widespread maternal malnutrition especially folic acid deficiency (Ahmed et al., 2024). Women in rural parts of Pakistan face additional risks due to limited access to prenatal care and nutritional guidance (Khan et al., 2023). The common risk factors overlapped between CLP populations as well as the need for local, region-specific research to learn exactly how these variables impact CLP in these populations.

Numerous studies have reported patterns including increased male incidence of CLP and a greater likelihood of clefts on the left side of the face (Lin et al., 2023). Apart from this, CLP usually comes with other health related problems such as congenital heart defects which also complicate the treatment and prognosis (Singh et al., 2024). One concerning issue in Pakistan is the delayed diagnosis and treatment, particularly in remote areas, leading to the children missing on the critical window for the early intervention (Javed et al., 2024). Other risk factors include family history of CLP (Lee et al., 2023), indicating a genetic predisposition and hence the need for genetic counseling in the at risk communities.

Ideally, the control of this possible future threat would be on a very local level, hence, there is a need for more detailed data about what is happening at that local level, and not statistics about the whole community. There is research on congenital anomalies at the national level, and yet there is no concentrated research on regions, for example, Southern Khyber Pakhtunkhwa (KPK), where the cultural practices and the access to the healthcare are not the same as the rest of the country (Iqbal et al, 2024). This gap is the focus of our study by examining incidence of CLP in Southern KPK along with the common factors such as consanguinity, maternal education, family history, and associated health problems. Such patterns offer a basis for public health interventions and can guide the design of culturally appropriate interventions.

This study, therefore, analyzes the data of 146 confirmed cases of CLP recorded over **7**-year period in two major tertiary hospitals of Southern KPK. Our research provides a comprehensive overview of the local landscape of CLP by looking at cleft types, laterality, age at diagnosis, maternal education, family demographics, etc. The purpose of these results is to strengthen the growing body of data about CLP in Pakistan and suggest actionable insights to healthcare providers and policymakers to collectively aid CLP prevention and management in Pakistan. The long term goal of this research is to contribute to better prevention, earlier detection, and improved outcomes for children born with cleft conditions in this region.

2. METHODOLOGY

In this study, we conducted a retrospective, a cross-sectional review at two major tertiary health care centers in southern Khyber Pakhtunkhwa (KPK), Pakistan. The two selected sites were Khalifa Gulnawaz Hospital, Bannu caring for patients in districts of Bannu, Kohat, Karak and North Waziristan and Mufti Mehmood Memorial Teaching Hospital Dera Ismail Khan caring for patients from the adjoining areas of districts of Dera Ismail Khan, South Waziristan, FR Dera Ismail Khan. Research period comprised 7-years period of July, 2017 to June, 2024. All the protocols of this study were prior approved by the appropriate ethical clearance from Institutional Review Board (IRB) of Gomal Medical College, MTI Dera Ismail Khan to ensure that the ethical guidelines for human participation in research were followed before starting the study.

In the study, a total of 223 clinically diagnosed cleft lip, cleft palate or both cleft lip and palate patients were included in the study. These cases were drawn from an initial screening of 1,230 patient records. Only those with complete, clearly documented history and demographic cases were included. Patients with ambiguous records or incomplete data were excluded to maintain the integrity of the analysis. Each patient's case was double-checked by two independent researchers to ensure accuracy and consistency of data extraction.

Data collection focused on several critical factors: patient demographics (such as age at the time of diagnosis and gender), specific type of cleft anomaly (cleft lip only, cleft palate only, or both), laterality (left-sided, right-sided, or bilateral), presence of any associated congenital anomalies, family history of similar conditions, parental consanguinity, and maternal details, including her socioeconomic factors, residence and education. Special emphasis was given on her education because of her awareness level and relationship to presentation of affected babies to hospitals for sake of correction procedures and treatment of relevant complications. Socioeconomic indicators and geographical background (rural or urban residence) were also recorded.

For data analysis, we used SPSS version 26.0. Descriptive statistics were applied to outline the frequency distribution of variables. To examine relationships between potential risk factors and the occurrence of CLP, chi-square tests were employed. For statistical significance, a p value of less than 0.05 was used, and if applicable, odds ratios with 95% confidence intervals were also determined to aid understanding the strength of associations.

Thus, the whole data entry and analysis process is cross checked independently so that the accuracy of data handling and observer bias are minimized. Pairwise deletion was used to handle missing values given that retaining missing values would not allow for compromising the analysis but would provide for the full utilization of available data.

Overall, this methodological approach makes an effort and analyses to overview the prevalence and risk factors of CLP in Southern KPK. The study focused on both clinical and socio demographic aspects with a view that the findings could support local healthcare planning and inform targeted preventive strategies.

3. RESULTS

In total, 223 medical records of two tertiary care centers in Southern Khyber Pakhtunkhwa, were reviewed, with an incidence rate of 1.83 per 1000 live births during the study period (July 2017 to June 2024). Below is a summary of the descriptive breakdown and statistical analysis of the findings.

1. Distribution of CLP Types

The most prevalent anomaly was cleft lip with palate (CLP), accounting for 46.6% (n =104) of total diagnosed cases. This was followed by isolated cleft lip in 34.9% (n = 78) and isolated cleft palate in 18.5% (n = 41). The distribution of types is presented in **Table 1** and visually illustrated in **Figure 1**.

Table 1: Distribution of CLP Types

Cleft Type	Frequency (n)	Percentage (%)
Cleft Lip with Palate	104	46.6%
Cleft Lip only	78	34.9%
Cleft Palate only	41	18.5%
Total	**223**	**100%**

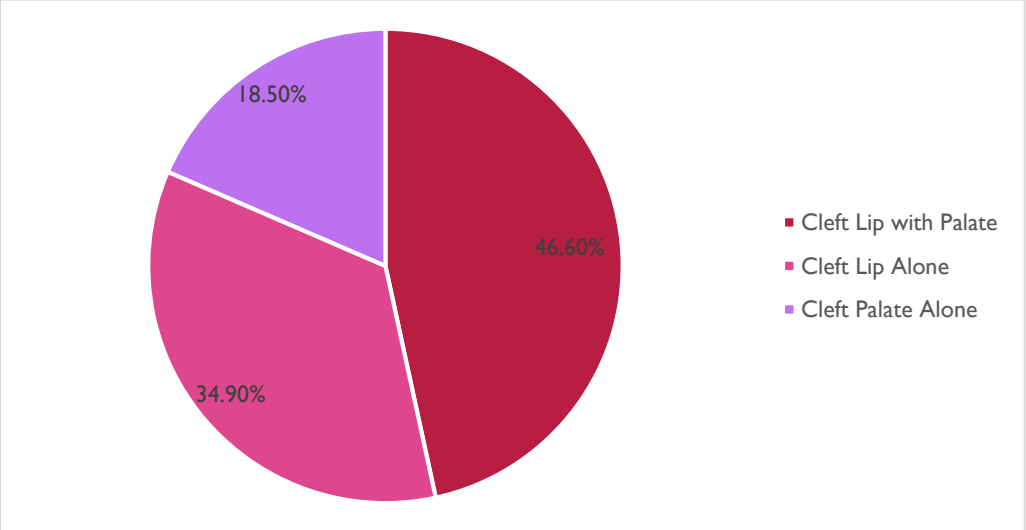

Figure 1: Pie chart showing proportions of cleft types.

2. Gender Distribution

A clear male predominance was observed, with 61.6% (n = 137) of patients being male and 38.4% (n = 86) female. The association between gender and CLP occurrence was statistically significant (p = 0.02). Detailed gender-wise distribution is shown in **Table 2** and visualized in **Figure 2**.

Table 2: Gender Distribution of CLP Cases

Gender	Frequency (n)	Percentage (%)	p-value
Male	137	61.6%	0.02
Female	86	38.4%	
Total	**223**	**100%**	

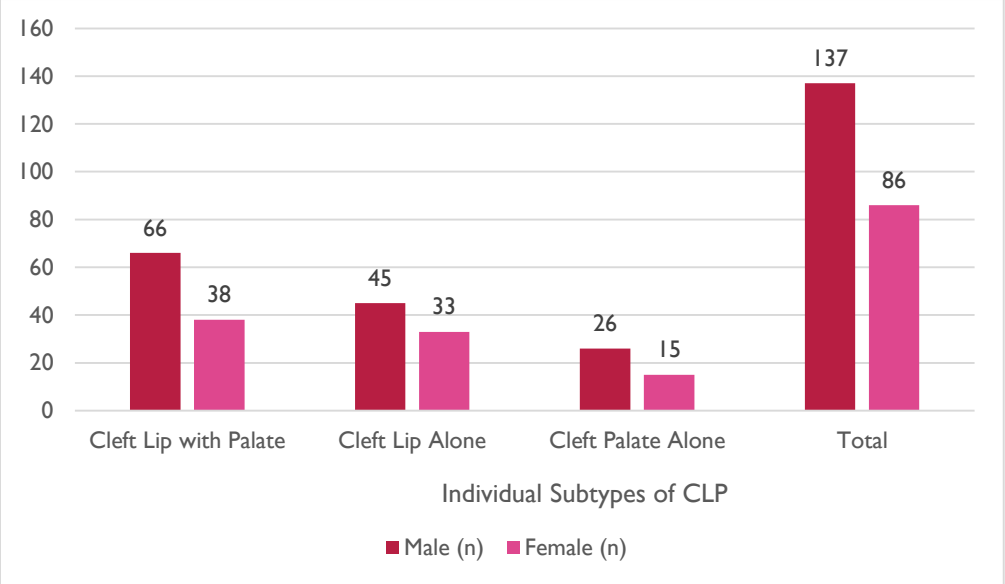

Figure 2: Bar chart comparing male and female distribution in different subtypes of CLP

3. Laterality of Clefts

Analysis of laterality showed that left-sided clefts were the most common, seen in 57.5% (n = 128) of cases. Right-sided clefts accounted for 28.8% (n = 64), while bilateral involvement was noted in 13.7% (n = 31). The difference in laterality was statistically significant (p = 0.04), as displayed in **Table 3** and **Figure 3**.

Table 3: Laterality of Clefts

Laterality	Frequency (n)	Percentage (%)
Left-sided	128	57.5%
Right-sided	64	28.8%
Bilateral	31	13.7%
Total	**223**	**100%**

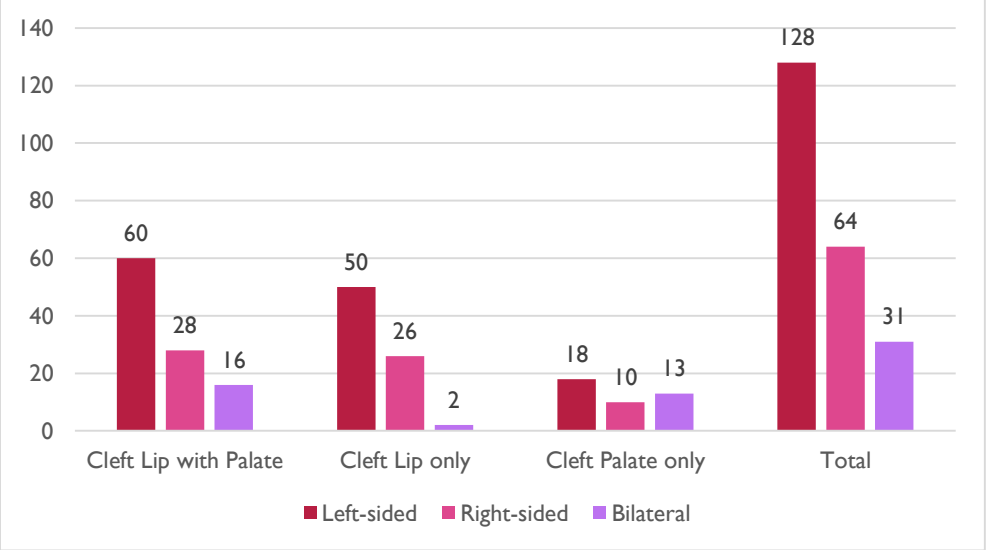

Figure 3: Bar graph depicting laterality patterns

4. Parental Consanguinity and Family History

Consanguineous marriages were reported in 83% (n = 185) of the cases, showing a significant correlation with CLP occurrence ($p < 0.0001$). A positive family history of CLP was present in 14.4% (n = 21) of patients ($p = 0.04$). These findings underscore the strong genetic component associated with CLP in the region. Details are provided in **Table 4**.

Table 4: Association of Consanguinity and Family History with CLP

Variable	Frequency (n)	Percentage (%)	p-value
Parental Consanguinity			
Present	185	83%	<0.0001
Absent	38	17%	
Family History			
Positive history	32	14.4%	0.04
No Family History	191	85.6%	

5. Maternal Education and Socioeconomic Factors

The data highlights the low educational attainment among mothers, with 72.6% having no formal education and only 4.8% achieving higher education. A statistically significant association ($p = 0.02$) was observed between maternal education and outcomes. The findings underscore the potential impact of maternal education and nutrition on health conditions. Table 5 represents these disparities through a table illustrating maternal education levels.

Table 5: Maternal Education

Parental Education Level	Frequency (n)	Percentage (%)
No formal education	162	72.6%
Primary education	32	14.4%
Secondary education	18	8.2%
Higher education	11	4.8%
Total	**223**	**100%**

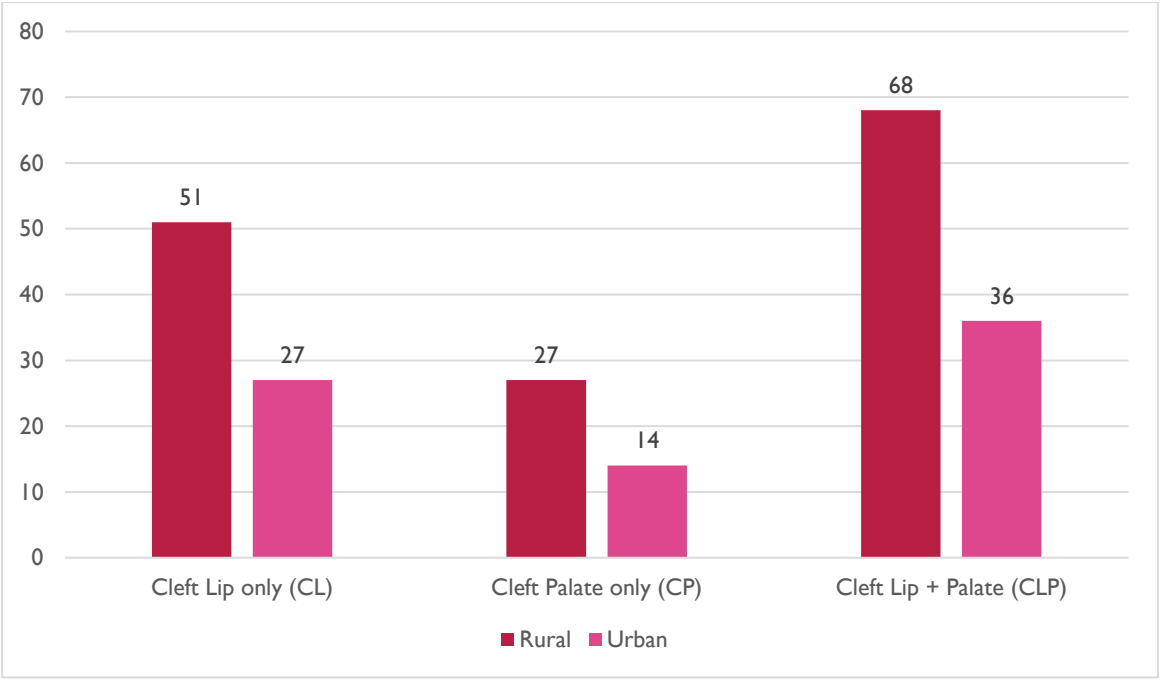

Figure 4: Bar graph showing parental socio-economic factors

6. Associated Congenital Anomalies and Age of Presentation

Among the CLP cases, associated congenital anomalies were documented in 21.2% (n = 47), with congenital heart defects being the most common anomaly. Regarding the age of presentation, 52.7% (n = 118) of patients presented within the first year of life. Presentation between one to two years accounted for 28.8% (n = 64), while delayed presentation beyond two years was observed in 18.5% (n = 41). These patterns are presented in **Table 6 and 7**.

Table 6: Associated Anomalies

Variable	Frequency (n)	Percentage (%)
Associated Congenital Anomalies	47	21.2%
No Associated Congenital Anomalies	176	78.8%
Associated Congenital Heart Defects	47	21.2%
Total	**223**	**100%**

Table 7: Age of Presentation of CLP Cases

Age Group at Presentation	Frequency (n)	Percentage (%)
≤ 1 year	118	52.7%
1–2 years	64	28.8%
> 2 years	41	18.5%
Total	**223**	**100%**

4. DISCUSSION

Our study provides valuable insights into the epidemiology of cleft lip and palate (CLP) in Southern Khyber Pakhtunkhwa, contributing to the understanding of genetic and environmental factors associated with orofacial clefts in this region. With a sample size of 223 patients collected over 7 years, our findings emphasize both the persisting burden of CLP and the multifactorial nature of its etiology.

One of the most striking observations was the high prevalence of parental consanguinity, noted in 83% of the cases. In line with this, is the role of consanguinity in being consistently associated with increased risk of congenital anomalies in the existing literature from similar regions with high consanguinity rates, such as rural Iran, and also parts of the Middle East (Rahimov et al., 2022; Yassin et al., 2023). The consanguineous marriages increase the frequency of homozygosity of the deleterious recessive allele and predisposing the offspring to congenital malformations such as CLP (Alade et al., 2023).

A large number of cases also originated from rural settings, which has previously been observed by studies conducted within sub–Saharan Africa and South Asia, where barriers to access to healthcare, as well as undernutrition, may act in concert with genetic risk (Mwanga et al., 2022; Singh et al., 2023). Barriers to prenatal care among both rural and tribal populations may delay or prevent diagnosis and modification of identified and non-identified risk factors during pregnancy (Harville et al., 2022).

Maternal education could not be separated from analysis. This bias was also observed among the majority of the mothers (72.6%) with no formal education further demonstrating the protective influence of education in maternal and child health outcomes. Poor health literacy (Kancheria el al., 2023; Zahid et al., 2024) and low levels of education tend to be associated with population that have poor access to prenatal supplements as well as suboptimal use of health services. Furthermore, the majority of the mothers (48.6%) are undernourished (especially low folate intake) which is known to be a risk factor in neural tube defects and orofacial clefts (Blencowe, et al. 2023). This is consistent with global evidence that maternal folic acid supplementation substantially reduces the risk of CLP (Zhou et al. 2023, Wilcox et al., 2022).

Our data seemed surprising in displaying a strong gender predilection, which has been noted in some previous reports with a male predominance in CLP cases (Mossey et al., 2022). Despite this, laterality patterns in our cohort mirrored those globally as left side clefts are more common, a phenomenon which in our embryology literature is not fully explained (Marazita, 2023).

Although no environmental or occupational exposure was reported to be associated with this deformity, family history became an important risk factor with 18.5% of the patients giving a positive family history of CLP. This adds additional genetic liability to CLP, as is supported by genome wide association studies identifying many regions involved in craniofacial development (Leslie et al., 2023, Yu et al., 2024).

Low income and limited access to healthcare further increased risk profile in our study population. This mirrors the conclusions of recent studies which also illustrate the overlap between poverty and congenital anomalies (e.g., Sitkin et al., 2023; Khan et al., 2023). The limited availability of specialized surgical and rehabilitative services in rural districts of Khyber Pakhtunkhwa aggravates the burden on affected families, prolonging functional and psychosocial challenges.

Our study's strength lies in its longitudinal design and comprehensive data collection from a region under-represented in global congenital anomaly research. However, limitations include its single-center scope and potential recall bias in parental reporting of nutritional intake and family history. Future research should aim for multi-centric studies with larger sample sizes and molecular genetic analyses to unravel the complex interplay of environmental and hereditary factors.

5. CONCLUSION

Our findings emphasize the need for targeted public health interventions, such as community education on consanguinity risks, improved maternal awareness nutrition programs, and enhanced access to prenatal care. Integrating genetic counseling and folic acid supplementation into routine maternal health services could substantially lessen the burden of CLP in high-risk populations. As global health priorities evolve, addressing the socio-genetic determinants of congenital anomalies must remain a focal point of maternal and child health strategies.

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