SHORT COMMUNICATION

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# Epidemiology of Craniofacial Anomalies: A Retrospective Analysis at a Tertiary Care Teaching Hospital in Southern India

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## ABSTRACT

Craniofacial anomalies are congenital malformations that impact essential functions like breathing, feeding, and speech, influenced by genetic and environmental factors. This retrospective study analyzed 54 cases of craniofacial anomalies in neonates at a tertiary care hospital in Karnataka, India, from 2022 to 2023. Data was extracted from electronic medical records and entered and analyzed using the Statistical Package for Social Sciences (SPSS) version 27.0. Most cases were from rural areas (92.6%) and predominantly male (68.5%), with 60.4% being first-born children. The notable percentage of CFAs among male, firstborn children from rural areas underscores the need for enhanced prenatal diagnostics and early intervention in underserved communities. Further research into region-specific genetic and environmental factors may provide valuable insights for reducing the incidence of CFAs.

**Keywords:** Craniofacial Anomalies, Cleft Lip, Cleft Palate, Microcephaly

## INTRODUCTION

Craniofacial anomalies (CFAs) encompass a broad and complex spectrum of congenital conditions. These anomalies can profoundly affect communication abilities, auditory function, facial structure, and cognitive development, resulting in lasting negative impacts on both health and social inclusion.[1] These anomalies result from disruptions in the development of neural crest cells, influenced by genetic and environmental


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factors.[2] A wide variety of congenital craniofacial anomalies exist, and the most prevalent among them is cleft lip and/or palate.[3]

In India, the reported prevalence of craniofacial anomalies is 1.1 per 1000 births.[4] A systematic review of congenital anomalies from India has shown that the prevalence of orofacial clefts is 15.69 per 10,000 live births.[5]

Considering the lack of studies in this region regarding the epidemiology of craniofacial anomalies, this study was planned to investigate the craniofacial anomalies and the factors associated with them.

## MATERIALS AND METHODS

The present retrospective study was conducted at a tertiary care teaching hospital in southern Karnataka. The study included neonates diagnosed with craniofacial anomalies who were born between October 2022 and February 2023. These neonates were either born in the hospital or referred to the hospital from other centers and were admitted for the management of craniofacial anomalies.

Approval of Institutional Ethical Committee: Kasturba Medical College and Kasturba Hospital Institutional Ethics Committee -2 (DHR Registration No: EC/NEW/INST/2021/1707), IEC2:37/2022 dated 10th Feb 2023.

The Institutional Ethics Committee (IEC) formally approved the study to ensure strict adherence to ethical standards. Data were collected retrospectively using the hospital's electronic medical records (EMR) system. A structured proforma was developed to extract relevant demographic and clinical data from the EMRs.

The data were entered into Microsoft Excel for data analysis. Results have been presented using frequency and percentage for categorical variables, and mean with standard deviation (SD) for continuous variables and Chi-square tests were conducted to examine the association between maternal and neonatal factors and the number of craniofacial anomalies. Wherever the assumptions for the Chi-square test were not met, Fisher's exact test was applied. $p<0.05$ is statistically significant.

## RESULTS

The study included 54 children born with craniofacial anomalies. The average age of the mothers was 29.2 years, and most families (92.6%) resided in rural areas. Regarding birth order, 60.4% of the children were firstborns, and 55.5% were delivered via cesarean section. Male children accounted for 68.5% of the cases, as shown in Table 1.

Among the anomalies, microcephaly was the most prevalent (46.2%), followed by cleft palate (31.4%) and cleft lip (18.5%). It was observed that 14.8% of the children had features suggestive of Down syndrome, as depicted in Table 2.

Based on the findings in Table 3, the analysis revealed patterns suggesting that multiple craniofacial anomalies were more frequently observed among firstborns, although these associations did not reach statistical significance.

**Table 1: Background characteristics of children born with craniofacial anomalies (n=54)**

| Characteristics | Cases (%) |
|---|---|
| **Age of mother (n=46)*** | 29.2 ± 4.5 |
| **Region (n=54)** | |
| Rural | 50(92.6) |
| Urban | 4(7.4) |
| **Order of pregnancy (n=48)** | |
| First childbirth | 29(60.4) |
| Second childbirth | 18(37.5) |
| Third childbirth | 1(2.1) |
| **Type of delivery (n=54)** | |
| Cesarean | 30(55.5) |
| Normal | 24(44.5) |
| **Gender of baby (n=54)** | |
| Male | 37(68.5) |
| Female | 17(31.5) |
| **Gestational age (n=53)** | |
| Full term | 32(60.4) |
| Premature | 16(30.2) |
| Post-term | 5(9.4) |
| Gestational age in weeks(n=53)* | 37(2.3) |
| Weight at birth in kgs (n=53)* | 2.7(0.9) |
| Height at birth in cm (n=49)* | 49.9(9.0) |
| Head circumference at birth in cm (n=50)* | 33.5(3.9) |

*Indicates Mean ± SD

**Table 2: Craniofacial anomalies observed in the children**

| Anomalies | Cases (%) |
|---|---|
| Microcephaly | 25(46.2) |
| Cleft palate | 17(31.4) |
| Cleft lip | 10(18.5) |
| Macrocephaly | 2(3.7) |
| Vascular malformation | 2(3.7) |
| Polydactyly | 2(3.7) |
| Open anterior fontanelle | 2(3.7) |
| Mild facial hypoplasia | 2(3.7) |
| Oxycephaly | 1(1.8) |
| Frontal bossing | 1(1.8) |
| Micrognathia | 1(1.8) |
| Left-sided facial hemangioma | 1(1.8) |
| Bilateral open lip schizencephaly | 1(1.8) |

Among the children, 8(14.8%) had features of down syndrome, Crouzon syndrome 1(1.8%) and Pierre robin syndrome 1(1.8%).

**Table 3: Association of Maternal and Neonatal Factors with the Craniofacial Anomalies**

| | Number of Craniofacial Anomalies | | p value |
|---|---|---|---|
| | More than one | One | |
| **Age of mother (n=46)** | | | |
| <35 years | 12(30.0) | 28(70) | 0.659# |
| ≥35 years | 1(16.7) | 5(83.3) | |
| **Region(n=54)** | | | |
| Rural | 14(28.0) | 36(72.0) | 0.573# |
| Urban | 2(50.0) | 2(50) | |
| **Oder of pregnancy(n=48)** | | | |
| First childbirth | 12(41.4) | 17(58.6) | |
| Second and third childbirth | 3(15.8) | 16(84.2) | 0.061 |
| **Gender of the baby (n=54)** | | | |
| Female | 6(35.3) | 11(64.7) | 0.537 |
| Male | 10(27.0) | 27(73.0) | |
| **Gestational age(n=53)** | | | |
| Full term | 10(31.3) | 22(68.8) | 0.822 |
| Post term | 1(20) | 4(80) | |
| Premature | 4(25) | 12(75) | |

# Indicates Fisher's exact test

## DISCUSSION

This retrospective study offers important insights into the characteristics of craniofacial anomalies (CFAs) among neonates in a tertiary care setting in southern India. Our findings indicate that microcephaly was the most common anomaly, consistent with observations from Junaid et al. who identified high rates of craniofacial malformations within a specific population.[6] Similarly, cleft palate and cleft lip had a common occurrence in our study after microcephaly, aligning with prior reports by Mishra et al. on the prominence of orofacial clefts in India.[7]

Our study revealed a clear male predominance (68.5%) in CFAs, echoing the findings by Odhiambo et al., who observed higher prevalence rates in male infants in Nairobi.[8] This gender disparity may suggest underlying biological influences on craniofacial development, though further research is needed to clarify these mechanisms. Additionally, our study noted that 60.4% of cases were firstborns, which aligns with patterns observed by Dufresne et al., who highlighted potential birth-order-related factors affecting CFA occurrence.[3]

Rural residency emerged as a significant characteristic, with 92.6% of the affected families residing in rural areas. This concentration could point to disparities in healthcare access, prenatal care, and genetic counseling. Shaw et al. emphasized the importance of equitable healthcare strategies in addressing such disparities, particularly in rural and underserved regions where CFAs may go undiagnosed or untreated.[1]

Notably, Down Syndrome was present in 14.8% of cases, a lower rate than syndromic craniosynostosis reported by Aljohar et al. in Saudi Arabia, where diverse regional factors likely influence diagnostic and prevalence rates.[9] Syndromic cases like Down Syndrome and other complex anomalies underscore the need for targeted interventions, as seen in studies by Sawasdipanich et al. which reported higher hospitalization rates for syndromic CFAs.[10]

Overall, the findings of our study underscore the need for improved access to early diagnostic resources, particularly in rural settings, to facilitate timely intervention for CFAs. Future studies focusing on regional genetic and environmental factors will be essential in understanding and mitigating the risks associated with CFAs.

## LIMITATIONS

Neonates with craniofacial anomalies often present with other systemic complications or require emergency admissions due to difficulties in breathing, respiratory distress, and associated malformations such as congenital heart disease, anomalies of the head and neck, and compromised central nervous systems. These additional complications are not highlighted in this study.

Due to the diverse nature of craniofacial anomalies and limited sample size, there is less description of the correlation between syndromic and non-syndromic craniofacial anomalies.

The research was carried out at a single institution with a relatively small sample size, which restricts the broader applicability of the results. Additionally, the study did not consider other systemic manifestations that are generally associated with syndromes.

## CONCLUSION

This study highlights a significant burden of craniofacial anomalies, with microcephaly and cleft palate being the most common types observed. The predominance of male infants and firstborns, along with the majority of cases originating from rural areas, underscores the importance of targeted healthcare strategies, including early detection and intervention in underserved populations. Future studies should aim to explore

genetic and environmental factors specific to this region, which could provide a deeper understanding of CFAs and inform more effective prevention and treatment efforts.

**Author contribution: AR** was involved in every aspect, including study conception, design, data collection, analysis, and manuscript preparation. **LU** and **SDM** contributed to most components, excluding data collection. **ATR** and **SS** played key roles in study conception, design, data collection, and manuscript preparation. **AK** and **LESL** were involved in the study's conception, design, and manuscript writing. All authors reviewed and approved the final manuscript, ensuring collective responsibility and intellectual contribution.

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