



CRANIOFACIAL ANOMALIES IN INDIA: A TERTIARY CARE-BASED STUDY

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Abstract

Introduction:

Craniofacial anomalies (CFAs) encompass a diverse group of congenital malformations affecting the skull, face, and associated structures, often resulting in functional and aesthetic impairments. In India, limited epidemiological data exists, particularly from tertiary care settings. This study aimed to evaluate the prevalence, demographic distribution, clinical characteristics, risk factors, and surgical outcomes of CFAs in a tertiary hospital in Delhi.

Materials and Methods: This study was conducted over 18 months at the Department of Burns, Plastic, and Maxillofacial Surgery, Safdarjung Hospital, New Delhi. A total of 112 patients with clinically diagnosed congenital CFAs were included. Patients with isolated cleft lip and/or cleft palate were excluded. Clinical, radiological, and surgical data were collected, and risk factors were assessed. Data analysis was performed using SPSS version 25.0.

Results: Most patients were aged 1–5 years (36.61%), and females predominated (64.3%). Non-syndromic CFAs were more common (54.5%) than syndromic cases (45.5%), with Apert syndrome being the most frequent syndromic diagnosis (52.9%). Facial involvement (56.3%) was more prevalent than cranial (25.9%) or combined involvement (17.9%). Among 44 craniosynostosis cases, scaphocephaly was the most common subtype. Isolated cleft palate (50.8%) was the leading facial anomaly. Improper iron and folic acid intake (16.96%) and family history (12.5%) were the most common risk factors. Multiple surgeries were required in 70.8% of surgically treated patients.

Conclusion: This study highlights the burden of CFAs in a tertiary care setting, emphasizing the predominance of non-syndromic cases and facial anomalies. Early diagnosis, nutritional interventions, genetic counselling, and multidisciplinary care are essential to improve outcomes and reduce long-term morbidity.

Keywords: Craniofacial anomalies, Congenital malformations, Craniosynostosis, Apert syndrome, Facial clefts, Tessier classification, Syndromic anomalies, Scaphocephaly, Multidisciplinary care, India, Paediatric surgery

Introduction

Craniofacial anomalies (CFAs) represent a broad spectrum of congenital structural malformations affecting the skull, face, and associated soft tissues, arising from disruptions in embryonic development. These anomalies range from minor cosmetic defects to severe impairments in breathing, feeding, speech, vision, and neurocognitive function [1]. Cleft lip and/or palate, the most common CFA, occurs in approximately 1 in 700 live births worldwide. However, despite this high prevalence, epidemiological data on CFAs in India, particularly from tertiary care centers, remain limited [2].

Although global initiatives aim to document congenital anomalies, data from many developing regions—including Asia and Africa—remain scarce [2]. Studies from the Middle East reveal varying prevalence: 1.9 per 1,000 live births in western Saudi Arabia and 7.98 per 1,000 pregnancies in Riyadh [3]. Other regional data report lower rates, including 0.3 per 1,000 in the UAE, and 1.5 and 2.4 per 1,000 live births in Oman and Jordan, respectively [3]. Craniosynostosis, involving premature cranial suture fusion, occurs in about 1 in 2,500 live births globally [4].

India lacks a centralized birth defect registry, leading to fragmented hospital-based data. Comprehensive studies are thus essential to assess prevalence, risk factors, and clinical patterns of CFAs in the Indian context. The etiology of CFAs is multifactorial, involving genetic factors such as chromosomal abnormalities, gene mutations, and disrupted developmental pathways, as well as environmental exposures including maternal nutritional deficiencies (notably folic acid), teratogenic drugs, infections, diabetes, and obesity [4–6]. Consanguinity—a known risk factor—remains prevalent in several Indian communities [7].

CFAs are commonly categorized into craniosynostosis and clefting syndromes. Craniosynostosis may be simple or compound, and either primary (genetic) or secondary (acquired) [8,9]. Syndromic forms, such as Apert, Crouzon, Pfeiffer syndrome, etc. are associated with systemic features [9]. Orofacial clefts follow Tessier's anatomical classification, ranging from midline to lateral facial clefts [10].

Advances in imaging, including high-resolution ultrasonography, fetal MRI, and postnatal CT/MRI, have improved diagnostic accuracy and surgical planning [11]. Management requires a multidisciplinary approach involving surgical reconstruction, orthodontics, speech therapy, and genetic counselling. Technologies such as 3D printing and CAD-based planning have enhanced surgical outcomes. However, in India, access to specialized care remains limited due to diagnostic delays, financial barriers, and lack of awareness [12].

This study was conducted to evaluate the prevalence, demographic profile, and clinical spectrum of CFAs in a tertiary care referral centre in India. It aims to identify common patterns, assess risk factors, and explore functional outcomes, thereby generating regional data to inform public health policy, early diagnosis, preventive strategies, and multidisciplinary management in the Indian healthcare setting.

Materials and Methods

This was a cross-sectional observational study conducted in the Department of Burns, Plastic, and Maxillofacial Surgery at Safdarjung Hospital, New Delhi. The study was conducted over a period of 18 months in 112 patients.

All patients presenting to the Department of Burns, Plastic, and Maxillofacial Surgery with a clinical diagnosis of congenital craniofacial anomalies were screened for eligibility. Patients of any age and gender were included, while those with isolated cleft lip and/or cleft palate were excluded. The study was approved by the Institutional Ethics Committee, and informed consent was obtained from the parents or guardians prior to clinical examination, radiological evaluation, and photography.

A comprehensive general physical examination was conducted, noting signs such as pallor, cyanosis, icterus, edema, clubbing, and lymphadenopathy. Vital signs and anthropometric parameters were recorded, including head circumference, interpupillary and interorbital distances, facial symmetry, and cephalic index. Systemic examination included cardiovascular, respiratory, and abdominal systems, along with neurological assessment. Associated anomalies such as syndactyly, anorectal malformations, or nail defects were also evaluated. Radiological evaluation included a non-contrast computed tomography (NCCT) scan of the head with 3D reconstruction to assess skull and facial bone anatomy.

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Analysis

All data were entered into Microsoft Excel and analyzed using SPSS software (version 25.0). Categorical variables were summarized using frequencies and percentages, while continuous variables were expressed as mean $\pm$ standard deviation or median with interquartile range (IQR), depending on data distribution. The Chi-square test or Fisher's exact test was used for comparing categorical variables. For continuous variables, the Student's t-test or Mann-Whitney U test was applied based on the normality of distribution. A p-value < 0.05 was considered statistically significant.

Results

A total of 112 patients with congenital craniofacial anomalies were enrolled in the study. The data were analyzed to determine the demographic profile, clinical classification, risk factors, and surgical management patterns. The majority of patients belonged to the 1–5 years age group (36.61%), followed by the 11–15 years (27.68%) and 6–10 years (23.21%) age groups. Only 12.5% of the cases were aged 16–20 years. (Table 1) Females predominated the study population, accounting for 64.3% of cases, while males constituted 35.7%. Non-syndromic craniofacial anomalies were slightly more common, representing 54.5% of the total, while 45.5% of patients had syndromic anomalies. Among the syndromic group, 52.9% were diagnosed with Apert syndrome (MC syndrome) (Table 2). Facial involvement alone was the most common presentation, observed in 56.3% of cases. Cranial involvement alone was noted in 25.9%, while 17.9% had both cranial and facial anomalies (Table 2). Of the 112 patients, 44 (39.3%) were diagnosed with craniosynostosis. Among these, non-syndromic craniosynostosis accounted for 65.91%, and syndromic craniosynostosis constituted 34.09%. Apert syndrome was the most prevalent syndromic association (15.91%), followed by Crouzon syndrome (6.82%), Treacher Collins syndrome (6.82%), and Goldenhar syndrome (4.55%). Scaphocephaly was the most frequent subtype ($n=19$), followed by plagiocephaly and mixed craniosynostosis ($n=8$ each). Lambdoid synostosis was observed in 6 cases, and metopic synostosis in 3. One case of cloverleaf skull (a subset of mixed craniosynostosis) was also reported. Among 63 patients with facial anomalies, isolated cleft palate was the most common presentation (50.8%), followed by combined cleft lip and palate (30.2%), and isolated cleft lip (19.0%). Syndactyly was the most frequent associated anomaly in syndromic patients, seen in 22 cases. Out of 109 patients with cleft classification data, midline clefts were most common (41.3%), followed by Tessier 1–13 clefts (16.5%), Tessier 2–12 (9.2%), and cleft 7 (8.3%). Other variants accounted for 21.1%. Rare anomalies like Sincipital encephalocele, Dandy Walker Malformation, Ventricular Cyst and White Tuft of hair were seen in one case each.

The most commonly identified risk factor was inadequate iron and folic acid supplementation during pregnancy (16.96%), followed by increased pain during gestation (15.18%), family history of craniofacial anomalies (12.5%), and antenatal polyhydramnios (7.14%). Only 1.79% of cases had a history of consanguinity. No patients reported teratogenic drug intake (Table 3). Out of 25 surgically managed patients, 70.8% required multiple surgeries, while 29.2% underwent a single surgery. A total of 9 surgeries were performed during the study period. Acrosyndactyly release for Apert syndrome was the most frequent procedure (6.25%), followed by fibrous dysplasia debulking (0.89%) and strip craniectomy for craniosynostosis (0.89%). Syndactyly release involved the feet in

3.57% and hands in 2.68% of patients. Fibrous dysplasia was reported in 2 patients (1.79%), highlighting its rare occurrence in this study.

Table 1. Demographic Distribution of Study Population

Variable	Category	Frequency (n)	Percentage (%)
Age Group (in years)	1–5	41	36.61%
	6–10	26	23.21%
	11–15	31	27.68%
	16–20	14	12.50%
Gender	Male	40	35.71%
	Female	72	64.29%

Table 2. Presentation of Craniofacial Anomalies

Variable	Category	Frequency (n)	Percentage (%)
Type of Anomaly	Cranial	29	25.9%
	Facial	63	56.3%
	Both	20	17.9%
Craniosynostosis Cases	Total	44	39.3%
	Non-Syndromic	29	65.91%
	Syndromic	15	34.09%
	• Apert Syndrome	7	15.91%
	• Crouzon Syndrome	3	6.82%
	• Treacher Collins Syndrome	3	6.82%
	• Goldenhar Syndrome	2	4.55%
	• Pfeiffer Syndrome	1	2.27%
	• Carpenter Syndrome	1	2.27%

Table 3. Risk Factors and Surgical Management

Variable	Category	Frequency (n)	Percentage (%)
Risk Factors	Improper IFA Intake	19	16.96%
	Increased Pain During Gestation	17	15.18%
	Family History of CFA	14	12.50%
	Antenatal Polyhydramnios	8	7.14%
	History of Consanguinity	2	1.79%
	Teratogenic Drug Intake	0	—
Surgeries Performed	Syndactyly Release (Apert)	7	6.25%
	• Feet	4	3.57%
	• Hand	3	2.68%
	Fibrous Dysplasia Debulking	1	0.89%
	Strip Craniectomy (Craniosynostosis)	1	0.89%
	Fronto-orbital advancement	1	0.87%
	Facial Bipartition	1	0.87%
Surgical Burden	Multiple Surgeries	22	70.8%
	Single Surgery	13	29.2%

Discussion

This study gives an evaluation of congenital craniofacial anomalies (CFAs) in a tertiary care setting, highlighting age and gender distribution, syndromic versus non-syndromic presentation, craniosynostosis patterns, facial cleft classifications, associated risk factors, and surgical interventions. In the present study, the majority of patients (36.61%) were within the 1–5 years age

group, followed by 27.68% in the 11–15 years category. This age distribution supports earlier detection of craniofacial anomalies, particularly those with visible facial involvement, and is consistent with previous findings by Aziza et al. (2011) [13], who observed that younger children constituted a significant proportion of craniofacial anomaly cases. Gender-wise, a female predominance (64.3%) was noted in contrast to Aziza et al. [13], who reported a higher incidence among males. This discrepancy may reflect regional, environmental, or referral biases and warrants further investigation. The current study reported a higher prevalence of non-syndromic cases (54.5%) compared to syndromic cases (45.5%). Among syndromic patients, Apert syndrome was the most frequent diagnosis (52.9%). These findings align with Suresh et al. (2023) [14] and Aziza et al. [13], the latter reporting that Apert and Crouzon syndromes were the most common syndromic craniosynostosis subtypes, comprising 38.5% and 27.7% of cases respectively. Our slightly higher Apert prevalence may indicate population-specific genetic predispositions or improved clinical recognition.

Facial involvement was the most commonly affected anatomical site (56.3%), followed by cranial anomalies (25.9%) and combined cranial-facial involvement (17.9%). These findings mirror the results of Aziza et al. [13], who found facial anomalies in 58.4% of their cohort. The greater visibility of facial deformities may facilitate earlier diagnosis and referral, while cranial anomalies, which often require imaging for detection, may be underreported in early infancy. Among 44 craniosynostosis cases, non-syndromic forms were predominant (65.91%). Apert syndrome (15.91%) was the leading syndromic subtype, followed by Crouzon (6.82%), Treacher Collins (6.82%), and Goldenhar syndromes (4.55%). These results correspond well with those of Aziza et al. [13], who documented a syndromic craniosynostosis rate of 43.6%. The Pongchangyoo et al. (2023) [15] study further supports the relevance of syndromic craniosynostosis by linking it to complications such as sleep-disordered breathing. Scaphocephaly emerged as the most frequent craniosynostosis subtype ($n = 19$), followed by plagiocephaly and mixed forms ($n = 8$ each), lambdoid synostosis ($n = 6$), and metopic synostosis ($n = 3$). This distribution is consistent with Hou et al. (2024) [16], who identified scaphocephaly as a predominant cranial deformity and emphasized the potential role of proteomic markers for early diagnosis. Aziza et al. [13] also observed a high incidence of scaphocephaly and stressed the importance of imaging and genetic screening. Cleft anomalies were predominantly represented by isolated cleft palate (50.8%), followed by cleft lip with palate (30.2%) and isolated cleft lip (19.0%). These results are consistent with both Aziza et al. [13], who reported cleft palate in 56.8% of cases, and Suresh et al. [14], who documented a high frequency of oral clefts in Indian children with congenital disorders. The predominance of cleft palate underscores the need for early feeding interventions and speech therapy. Among syndromic patients, syndactyly was the most common associated anomaly ($n = 22$), a hallmark feature of Apert syndrome. This finding corresponds with earlier data from Aziza et al. [13] and Patel et al. (2022) [17], who noted high rates of musculoskeletal anomalies in syndromic cases. Syndactyly's frequent co-occurrence with craniofacial anomalies necessitates timely orthopedic intervention alongside craniofacial care. Tessier cleft classification analysis revealed a predominance of midline clefts (41.3%), followed by Tessier 1–13 (16.5%), Tessier 2–12 (9.2%), and Cleft 7 (8.3%). This is in agreement with Trindade et al. (2006) [18] and Obuekwe & Ojo (2020) [19], both of whom identified midline clefts as the most frequent in their respective studies. Interestingly, Kim et al. (2020) [20] found a higher prevalence of Cleft 7 in Korean patients, illustrating how regional or ethnic variation influences cleft pattern distribution. Our findings reinforce the complexity of midline clefts and the necessity of a multidisciplinary approach to their management. Risk factor analysis highlighted improper maternal iron and folic acid intake (16.96%) as the leading factor, followed by increased gestational pain (15.18%), family history of craniofacial anomalies (12.5%), and polyhydramnios (7.14%). Only 1.79% of patients reported consanguinity. These observations align with Aziza et al. [13] and Patel et al. [17], both of whom found significant associations between maternal nutritional deficiencies, consanguinity, and congenital anomaly prevalence. P.N. et al. (2023) [21] emphasized prenatal screening and maternal counselling as key preventive strategies—a conclusion further similar to our findings. Regarding surgical intervention, 70.8% of

patients underwent multiple procedures, while 29.2% had a single surgery. This reflects the complex nature of CFAs, which often necessitate staged or multi-specialty intervention. Morris et al. (2024) [22] and Noad et al. (2024) [23] reported similar long-term surgical burdens in congenital anomaly patients, emphasizing the need of follow-up protocols. Nine surgeries were performed during the study period, with syndactyly release being the most common (6.25%), followed by debulking for fibrous dysplasia and strip craniectomy (each 0.89%). These figures align with Allareddy et al. (2015) [24], who documented high rates of orthopaedic and craniofacial surgeries in affected children. Aziza et al. [13] similarly noted that most craniofacial anomaly cases required multiple surgical interventions, underscoring the need for integrated, long-term care.

Conclusion

This study highlights the clinical and demographic profile of congenital craniofacial anomalies in a tertiary care setting, with non-syndromic cases and facial involvement being most common. Apert syndrome was the predominant syndromic condition, often requiring multiple surgeries. Scaphocephaly was the most frequent craniosynostosis subtype. Key risk factors included inadequate maternal iron and folic acid intake and family history. The findings underscore the need for early diagnosis, genetic counselling, and multidisciplinary care to improve outcomes and reduce the burden of craniofacial anomalies.

Limitations

This single-center study may not reflect the broader population. The sample size was limited by case availability, and exclusion of isolated cleft lip and palate may have underrepresented the full spectrum of anomalies. Systemic associations and genetic testing and long-term surgical outcomes were not comprehensively assessed.

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