








# Epidemiology and Multidisciplinary Approach in Orofacial Clefts: Clinical Features and Rehabilitation Patterns

## *Epidemiologia e abordagem multidisciplinar nas fissuras orofaciais: Características clínicas e padrões de reabilitação*

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### Abstract

#### Keywords

- cleft lip
- cleft palate
- craniofacial abnormalities
- epidemiology
- surgery, plastic

**Introduction** Orofacial clefts are congenital malformations that affect craniofacial development and represent a significant public health concern. The present study aimed to investigate the frequency, distribution, and characteristics of these clefts in patients treated at a specialized center, also evaluating the rehabilitation patterns employed.

**Materials and Methods** The research involved 59 patients diagnosed with different types of orofacial clefts, including both simple and complex cases, with or without surgical history. We analyzed variables such as sex, age group, and procedures performed. The statistical analysis was conducted using multivariate methods, including principal component analysis (PCA) and cluster analysis (CA), with the SAS (SAS Institute Inc.) software, version 9.2.

**Results** Cleft right (CRT) or left transforamen (CLT) were the most prevalent, particularly among male patients. Most cases involved infants and young children, reflecting the practice of early intervention. Approximately 59.3% of the patients had

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already undergone surgeries such as cheiloplasty and palatoplasty, often in combination. More complex cases, such as those of cleft bilateral transforaminal (CBT), required multiple surgeries and extended follow-up.

**Conclusion** The current study highlights the need for early and continuous interventions, as well as regional epidemiological surveys to guide public policies and optimize resource allocation. Multidisciplinary care and personalized treatment planning are essential to improving the quality of life of the patients and their families.

## Resumo

**Introdução** As fissuras orofaciais são malformações congênitas que afetam o desenvolvimento craniofacial e configuram um problema relevante de saúde pública. Este estudo buscou investigar a frequência, a distribuição e as características dessas fissuras em pacientes atendidos em um centro especializado, além de avaliar os padrões de reabilitação adotados.

**Materiais e Métodos** A pesquisa envolveu 59 pacientes diagnosticados com diferentes tipos de fissuras orofaciais, incluindo casos simples e complexos, com ou sem histórico cirúrgico. Foram analisadas variáveis como gênero, faixa etária e procedimentos realizados. A análise estatística foi realizada por métodos multivariados, incluindo análise de componentes principais (PCA, do inglês *principal component analysis*) e análise de agrupamento (CA, do inglês *cluster analysis*), com o programa SAS (SAS Institute Inc.), versão 9.2.

**Resultados** As fissuras transforaminais direitas (CRT, do inglês *cleft right transforamen*) ou esquerdas (CLT, do inglês *cleft left transforamen*) foram as mais prevalentes, especialmente em pacientes do sexo masculino. A maior parte dos atendimentos foi feita em bebês e crianças pequenas, o que reflete a prática de intervenção precoce. Cerca de 59,3% dos pacientes já haviam passado por cirurgias, como queiloplastia e palatoplastia, muitas vezes em combinação. Fissuras mais complexas, como as transforaminais bilaterais (CBT, do inglês *cleft bilateral transforaminal*), demandaram múltiplas cirurgias e acompanhamento prolongado.

**Conclusão** Este estudo ressalta a necessidade de intervenções precoces e contínuas, bem como de levantamentos epidemiológicos regionais para orientar políticas públicas e otimizar a alocação de recursos. O manejo multidisciplinar e o planejamento personalizado são fundamentais para melhorar a qualidade de vida dos pacientes e de suas famílias.

## Palavras-chave

- anormalidades craniofaciais
- cirurgia plástica
- epidemiologia
- fenda labial
- fissura palatina

## Introduction

Orofacial clefts are congenital malformations that impair craniofacial development and represent a significant public health issue in various regions worldwide. The prevalence of these conditions is estimated to be of approximately 1 in every 700 live births, underscoring their clinical and epidemiological importance.<sup>1</sup> Between 1990 and 2019, the global incidence of these clefts declined, particularly in regions with high sociodemographic index (SDI) levels. However, some low-income countries have reported significant increases in occurrence rates.<sup>2</sup>

Orofacial clefts result from a combination of genetic and environmental factors. The genetic factors include mutations in genes that regulate facial and palate development. Additionally, exposure to certain medications and chemicals during pregnancy can interfere with fetal develop-

ment, increasing the risk of these malformations.<sup>3</sup> There is also evidence that maternal nutrition plays a crucial role in the etiology of cleft lip and palate, along with epigenetic modifications that contribute to its pathogenesis.<sup>4,5</sup>

These clefts range from isolated lip clefts to complete palatal involvement, impacting facial esthetics as well as essential functions such as feeding, speech, and breathing. These impairments significantly affect the quality of life of the patients and their families.<sup>6,7</sup>

Given the complexity of these conditions, managing orofacial clefts requires a multidisciplinary approach involving surgeons, speech therapists, orthodontists, psychologists, and other healthcare professionals.<sup>8</sup> Early intervention is crucial to optimize the long-term outcomes, as procedures such as cheiloplasty and palatoplasty, performed in early childhood, minimize functional difficulties and facilitate

psychosocial development.<sup>9</sup> However, the need for multiple surgeries over time makes continuous follow-up essential to ensure effective rehabilitation.<sup>10</sup>

The epidemiology of orofacial clefts varies according to genetic, environmental, and regional factors, making it necessary to map these variables to guide more effective interventions.<sup>11</sup> Although significant advances have been made in diagnosis and treatment, gaps remain in the literature, particularly regarding the distribution of cleft types and the impact of surgical history on outcomes across different populations.<sup>12</sup> Such information is critical to optimize resource allocation and improve the quality of care provided by specialized centers.

## Objective

The present study aims to investigate the frequency, distribution, and characteristics of orofacial clefts in patients treated at Pró-Sorriso Center for Cleft Lip and Palate Treatment, located in the city of Alfenas, state of Minas Gerais, Southeastern Brazil. Furthermore, it seeks to assess the rehabilitation patterns employed, considering variables such as sex, age, and surgical history, with the goal of contributing to the development of more effective treatment strategies and public policies focused on patient rehabilitation.

## Materials and Methods

The current study included 59 patients with orofacial clefts, with or without a clinical history of orofacial surgery, treated between February 2019 and February 2020. The patients' ages ranged from 3 months to 58 years (mean:  $10.1 \pm 14.6$  years), encompassing both sexes (17 female subjects, with a

mean age of  $16.3 \pm 19.3$  years; and 42 male subjects, with a mean age of  $7.6 \pm 11.6$  years). All patients were clinically indicated for surgical rehabilitation and were diagnosed according to the clinical classification of orofacial clefts.<sup>13,14</sup> They received continuous medical and dental care at Pró-Sorriso Center.

The patient population was stratified by age group, sex, type of orofacial cleft, surgical history, and type of previous surgical rehabilitation. These variables were statistically analyzed to assess trends and associations. The present research adhered to the ethical principles outlined in Resolution no. 466/2012 of the Brazilian National Health Council and was approved by the Research Ethics Committee of Faculdade de Odontologia de Piracicaba, Universidade Estadual de Campinas (FOP/UNICAMP), under protocol No. 093/2014, CAAE 34875614.0.0000.5418.

Data were subjected to multivariate statistical analysis using the SAS (SAS Institute Inc.) software, version 9.2. The analytical methods included principal component analysis (PCA) and interactive biplot construction, along with cluster analysis (CA) and dendrogram interpretation (threshold  $\leq 0.5$ ) to identify patterns and relationships within the data.<sup>15</sup>

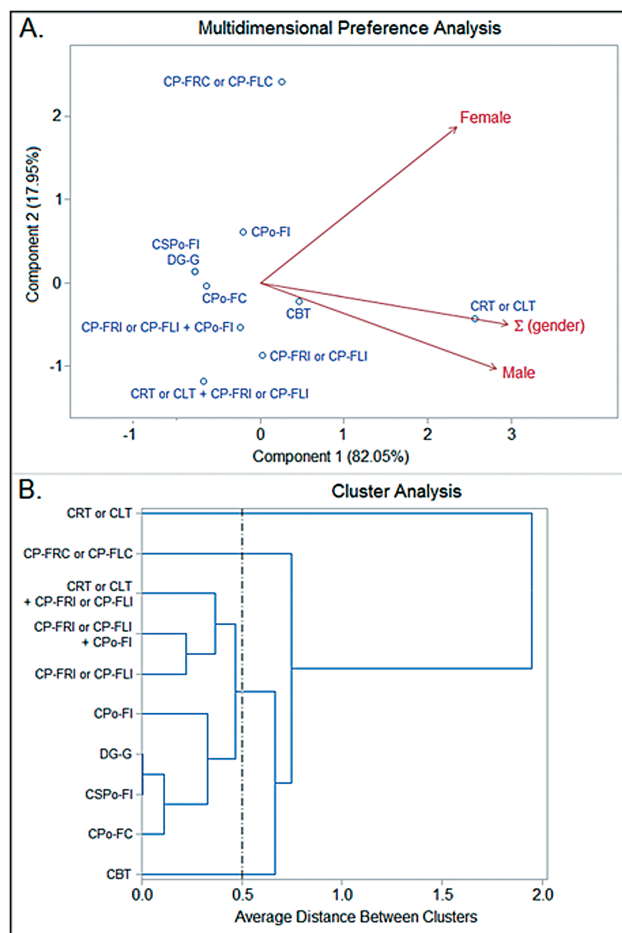
## Results

The present study analyzed 59 patients diagnosed with various types of orofacial clefts, encompassing anatomical variations and associated syndromic conditions, as shown in ►Table 1 and ►Fig. 1. A clear predominance of transforamen clefts (cleft right transforamen [CRT] and cleft left transforamen [CLT]) was observed, with a higher incidence among male patients, reflecting an epidemiological profile characteristic of these malformations.

**Table 1** Epidemiological characteristics of the patients with orofacial clefts, detailing the incidence by sex and cleft type.

Types of orofacial clefts	Female		Male		$\Sigma$	
	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%
CBT	2	3.4	7	11.9	9	15.3
CRT or CLT	4	6.8	18	30.5	22	37.3
CPo-FC	1	1.7	1	1.7	2	3.4
CPo-FI	2	3.4	2	3.4	4	6.8
CP-FRC or CP-FLC	4	6.8	1	1.7	5	8.5
CP-FRI or CP-FLI	1	1.7	6	10.2	7	11.9
CSPo-FI	1	1.7	0	0.0	1	1.7
DG-G	1	1.7	0	0.0	1	1.7
CP-FRI or CP-FLI + CPo-FI	1	1.7	4	6.8	5	8.5
CRT or CLT + CP-FRI or CP-FLI	0	0.0	3	5.1	3	5.1
$\Sigma$	17	28.8	42	71.2	59	100.0

**Abbreviations:** CBT, cleft bilateral transforaminal; CLT, cleft left transforamen; CP-FLC, cleft preforamen left complete; CP-FLI, cleft preforamen left incomplete; CP-FRC, cleft preforamen right complete; CP-FRI, cleft preforamen right incomplete; CPo-FC, cleft postforamen complete; CPo-FI, cleft postforamen incomplete; CRT, cleft right transforamen; CSPo-FI, cleft submucosa postforamen incomplete; DG-G, deformity groove-gingival.



**Fig. 1** Biplot analyses using principal component analysis (PCA) and dendrograms from cluster analysis (CA) based on the frequency distribution of the different types of orofacial clefts and their epidemiological variable (sex). Orofacial clefts: 1) cleft bilateral transforaminal (CBT); 2) cleft right transforamen (CRT) or cleft left transforamen (CLT); 3) cleft postforamen complete (CPo-FC); 4) cleft postforamen incomplete (CPo-FI); 5) cleft preforamen right complete (CP-FRC) or cleft preforamen left complete (CP-FLC); 6) cleft preforamen right incomplete (CP-FRI) or cleft preforamen left incomplete (CP-FLI); 7) cleft submucosa postforamen incomplete (CSPo-FI); 8) deformity groove-gingival (DG-G); 9) CP-FRI or CP-FLI + CPo-FI; and 10) CRT or CLT + CP-FRI or CP-FLI.

The findings revealed that males were more frequently affected by CRT and CLT. In contrast, more complex clefts, such as cleft preforamen complete (CP-FC) or cleft preforamen incomplete (CP-FI), exhibited a more balanced distribution across both sexes. This suggests potential variations in the etiological or environmental factors influencing different cleft types. The stratification by sex and cleft type provides insights that may assist in tailoring clinical interventions according to demographic patterns.

Additionally, patients with clefts associated with other malformations (such as a combination of trans- and preforamen clefts) exhibited a notable distribution across age groups, as shown in ▶Table 2 and ▶Fig. 2. The analysis of age groups revealed that infants (up to 1 year old) and

toddlers (1–3 years) were most frequently hospitalized for initial surgical interventions, indicating an established practice of early intervention. These interventions aim to minimize functional and esthetic impairments, promoting social and psychosocial development.

Approximately 59.3% of the patients, as shown in ▶Table 3 and ▶Fig. 3, had undergone at least 1 surgical intervention. The most common procedures included cheiloplasty, palatoplasty, and combined surgeries, such as cheiloplasty with palatoplasty. More complex cases, such as those of cleft bilateral transforaminal (CBT), required multiple surgeries over time, underscoring the importance of long-term planning and continuous follow-up to ensure effective rehabilitation.

The multivariate analysis, performed using the SAS software, version 9.2, enabled the construction of biplots and dendrograms to interpret clustering patterns and similarities between cleft types and their epidemiological characteristics. ▶Table 4 and ▶Fig. 4 highlight the relationship between cleft types and the surgical interventions performed. We observed that patients with simpler clefts, such as CRT or CLT, often underwent isolated cheiloplasty or a combination of cheiloplasty and palatoplasty. In contrast, more complex cases, such as those of clefts associated with other malformations, required additional procedures, including rhinoseptoplasty.

## Discussion

The predominance of CLT over CRT is a striking trend in the epidemiology of lip and palate clefts, particularly among male patients. Studies<sup>16</sup> indicate that males are significantly more affected by these clefts, with a male-to-female ratio of 1.34:1. In a cohort from West Bengal,<sup>16</sup> 69.59% of the patients were male, reinforcing this sex disparity.

The left side of the face is more frequently affected, with left unilateral clefts being the most common, as observed in several studies.<sup>17,18</sup> In Brazil, Rollemberg et al.<sup>19</sup> (2019) found that 20.5% of patients presented with CLT, supporting this trend.

The high frequency of CRT and CLT highlights the importance of early surgical interventions to ensure proper orofacial function, such as feeding and speech.<sup>18</sup> More complex clefts, such as CBT and cleft postforamen incomplete (CPo-FI), exhibit a more balanced sex distribution, contrasting with the male predominance in other types.<sup>20</sup> These complex clefts require more elaborate surgical approaches and long-term multidisciplinary planning.<sup>21</sup>

The success of interventions also depends on continuous follow-up, as multiple surgeries may be necessary throughout the patient's life.<sup>10</sup> In this context, advanced diagnostic tools can enhance early detection and management, positively impacting therapeutic outcomes.<sup>22</sup> Additionally, it is crucial to consider the psychological and socioeconomic impacts on both patients and their families.<sup>4</sup>

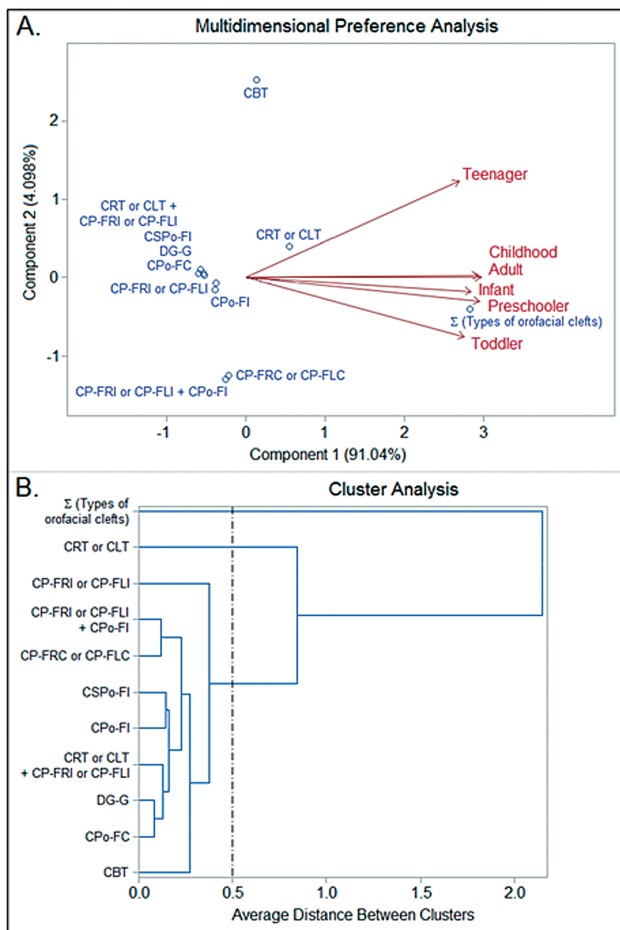
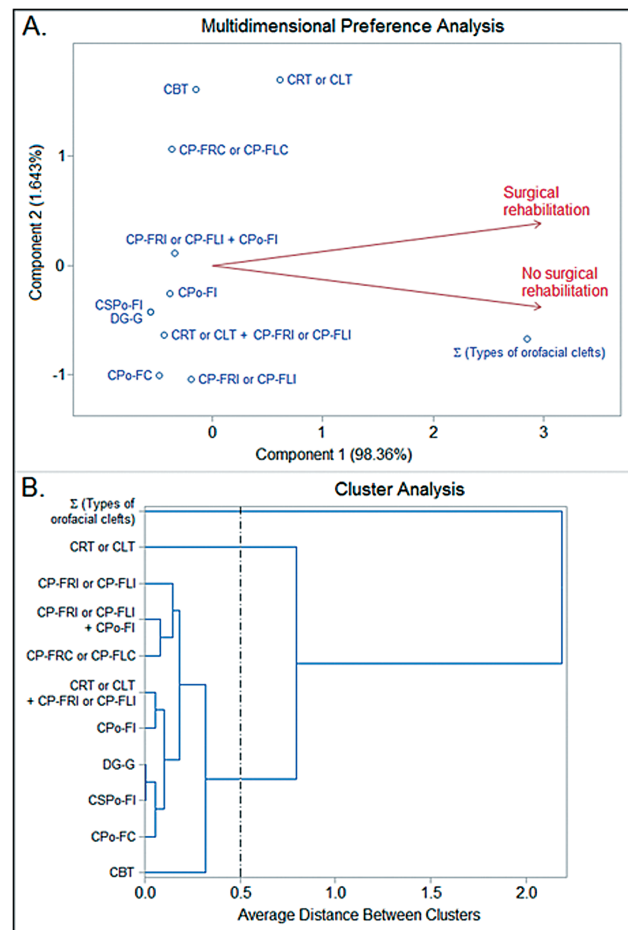
In the current study, 59.3% of patients had undergone some form of surgical intervention, with cheiloplasty and palatoplasty being the most frequent. Cheiloplasty,

**Table 2** Epidemiological characteristics of the patients with orofacial clefts according to age group and cleft type

Types of orofacial clefts	Infant		Toddler		Pre-schooler		Childhood		Teen-ager		Adult		$\Sigma$	
	n	%	n	%	n	%	n	%	n	%	n	%	n	%
CBT	1	1.7	1	1.7	1	1.7	2	3.4	2	3.4	2	3.4	9	15.3
CRT or CLT	10	16.9	1	1.7	2	3.4	4	6.8	1	1.7	4	6.8	22	37.3
CPo-FC	1	1.7	0	0.0	0	0.0	0	0.0	0	0.0	1	1.7	2	3.4
CPo-FI	1	1.7	0	0.0	1	1.7	2	3.4	0	0.0	0	0.0	4	6.8
CP-FRC or CP-FLC	0	0.0	2	3.4	1	1.7	1	1.7	0	0.0	1	1.7	5	8.5
CP-FRI or CP-FLI	5	8.5	0	0.0	0	0.0	1	1.7	0	0.0	1	1.7	7	11.9
CSPo-FI	0	0.0	0	0.0	0	0.0	1	1.7	0	0.0	0	0.0	1	1.7
DG-G	1	1.7	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	1	1.7
CP-FRI or CP-FLI + CPo-FI	1	1.7	2	3.4	1	1.7	0	0.0	0	0.0	1	1.7	5	8.5
CRT or CLT + CP-FRI or CP-FLI	2	3.4	0	0.0	0	0.0	1	1.7	0	0.0	0	0.0	3	5.1
$\Sigma$	22	37.3	6	10.2	6	10.2	12	20.3	3	5.1	10	16.9	59	100.0

**Abbreviations:** CBT, cleft bilateral transforaminal; CLT, cleft left transforamen; CP-FLC, cleft preforamen left complete; CP-FLI, cleft preforamen left incomplete; CP-FRC, cleft preforamen right complete; CP-FRI, cleft preforamen right incomplete; CPo-FC, cleft postforamen complete; CPo-FI, cleft postforamen incomplete; CRT, cleft right transforamen; CSPo-FI, cleft submucosa postforamen incomplete; DG-G, deformity groove-gingival.

**Notes:** Infant (from birth to 1 year of age); toddler (1–3 years of age); preschooler (3–5 years of age); childhood (6–11 years of age); teenager (12–17 years of age); and adult (older than 18 years).

**Fig. 2** Biplot analyses using PCA and dendrograms from CA based on the frequency distribution of the different types of orofacial clefts and their epidemiological variable (age group).**Fig. 3** Biplot analyses using PCA and dendrograms from CA based on the frequency distribution of the different types of orofacial clefts and their epidemiological variable (surgical history).

**Table 3** Epidemiological characteristics of the patients with orofacial clefts based on surgical history and cleft type

Types of orofacial clefts	Surgical rehabilitation		No surgical rehabilitation		$\Sigma$	
	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%
CBT	7	11.9	2	3.4	9	15.3
CRT or CLT	15	25.4	7	11.9	22	37.3
CPo-FC	0	0.0	2	3.4	2	3.4
CPo-FI	2	3.4	2	3.4	4	6.8
CP-FRC or CP-FLC	4	6.8	1	1.7	5	8.5
CP-FRI or CP-FLI	3	5.1	4	6.8	7	11.9
CSPo-FI	0	0.0	1	1.7	1	1.7
DG-G	0	0.0	1	1.7	1	1.7
CP-FRI or CP-FLI + CPo-FI	3	5.1	2	3.4	5	8.5
CRT or CLT + CP-FRI or CP-FLI	1	1.7	2	3.4	3	5.1
$\Sigma$	35	59.3	24	40.7	59	100.0

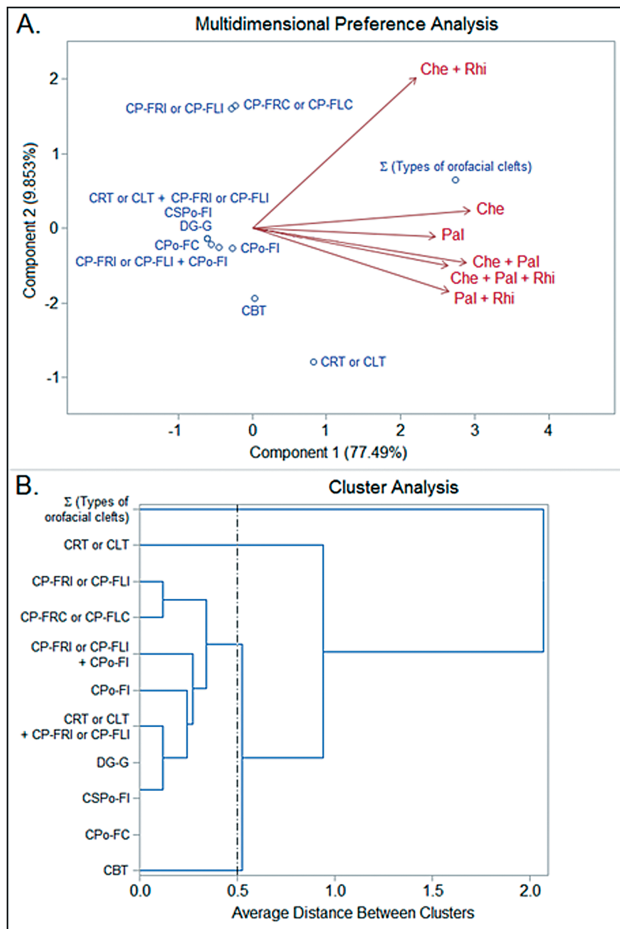
**Abbreviations:** CBT, cleft bilateral transforaminal; CLT, cleft left transforamen; CP-FLC, cleft preforamen left complete; CP-FLI, cleft preforamen left incomplete; CP-FRC, cleft preforamen right complete; CP-FRI, cleft preforamen right incomplete; CPo-FC, cleft postforamen complete; CPo-FI, cleft postforamen incomplete; CRT, cleft right transforamen; CSPo-FI, cleft submucosa postforamen incomplete; DG-G, deformity groove-gingival.

**Table 4** Epidemiological characteristics of the patients with orofacial clefts according to the incidence of previous surgical rehabilitation types and cleft type

Types of orofacial clefts	Surgical rehabilitation																No surgical rehabilitation		Σ	
	Che		Pal		Rhi		Che + Pal		Che + Rhi		Pal + Rhi		Che + Pal + Rhi		Σ					
	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%	<i>n</i>	%
CBT	1	1.7	0	0.0	0	0.0	4	6.8	0	0.0	0	0.0	2	3.4	7	11.9	2	3.4	9	15.3
CRT or CLT	7	11.9	1	1.7	0	0.0	5	8.5	0	0.0	1	1.7	1	1.7	15	25.4	7	11.9	22	37.3
CPo-FC	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	2	3.4	2	3.4
CPo-FI	0	0.0	2	3.4	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	2	3.4	2	3.4	4	6.8
CP-FRC or CP-FLC	3	5.1	0	0.0	0	0.0	0	0.0	1	1.7	0	0.0	0	0.0	4	6.8	1	1.7	5	8.5
CP-FRI or CP-FLI	2	3.4	0	0.0	0	0.0	0	0.0	1	1.7	0	0.0	0	0.0	3	5.1	4	6.8	7	11.9
CSPo-FI	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	1	1.7	1	1.7
DG-G	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0	1	1.7	1	1.7
CP-FRI or CP-FLI + CPo-FI	1	1.7	0	0.0	0	0.0	2	3.4	0	0.0	0	0.0	0	0.0	3	5.1	2	3.4	5	8.5
CRT or CLT + CP-FRI or CP-FLI	0	0.0	0	0.0	0	0.0	1	1.7	0	0.0	0	0.0	0	0.0	1	1.7	2	3.4	3	5.1
Σ	14	23.7	3	5.1	0	0.0	12	20.3	2	3.4	1	1.7	3	5.1	35	59.3	24	40.7	59	100.0

**Abbreviations:** CBT, cleft bilateral transforaminal; Che, cheiloplasty; CLT, cleft left transforamen; CP-FLC, cleft preforamen left complete; CP-FLI, cleft preforamen left incomplete; CP-FRC, cleft preforamen right complete; CP-FRI, cleft preforamen right incomplete; CPo-FC, cleft postforamen complete; CPo-FI, cleft postforamen incomplete; CRT, cleft right transforamen; CSPo-FI, cleft submucosa postforamen incomplete; DG-G, deformity groove-gingival; Pal, palatoplasty; Rhi, rhinoseptoplasty.





**Fig. 4** Biplot analyses using PCA and dendrograms from CA based on the frequency distribution of the different types of orofacial clefts and their epidemiological variable (types of previous surgical rehabilitation). **Abbreviations:** Che, Cheiloplasty; Pal, palatoplasty; Rhi, rhinoseptoplasty.

performed between 3 and 6 months of age, aims to restore lip structure and function by aligning its components and reestablishing muscular continuity.<sup>23,24</sup> Palatoplasty, typically performed around 10 months of age, seeks to restore palatal integrity using techniques such as intravelar veloplasty and vomer flap closure.<sup>23,25</sup> These procedures are fundamental in restoring both functionality and esthetics, promoting psychosocial development.<sup>6</sup>

Beyond improving facial appearance, these surgeries enhance speech and feeding capabilities, which are essential for social integration and quality of life.<sup>14,26</sup> Studies<sup>27</sup> suggest that the timing and techniques of interventions can significantly impact long-term outcomes.

Personalizing treatment plans is essential, particularly in more complex cases, such as CRT or CLT combined with other anomalies. A quantified classification of clefts assists in planning individualized strategies, ensuring greater treatment efficacy.<sup>28</sup> Patients with seemingly similar conditions may respond differently to interventions, underscoring the need for personalized approaches.<sup>29</sup>

A study<sup>30</sup> involving 2,475 patients found that comorbidities often require more comprehensive rehabilitation, highlighting the need for continuous specialized care. Multidisciplinary management is essential to achieve satisfactory outcomes, as the lack of integration among specialties can compromise treatment effectiveness.<sup>10</sup> While individualized treatment is a priority, it is also important to address the risk of patient fatigue, which may affect adherence and long-term therapeutic success.

The multidisciplinary approach, often emphasized in the literature,<sup>31</sup> is indispensable for effective rehabilitation. Collaboration among speech therapists, psychologists, and orthodontists is crucial to ensure that functional, emotional, and social aspects are comprehensively addressed.<sup>9</sup> This holistic approach is essential to promote psychosocial development and enhance patient quality of life.<sup>32</sup>

Finally, the current study highlights the importance of regional epidemiological surveys as a basis for the development of more effective public policies. Analyzing the distribution of cleft types and the impact of surgical interventions provides valuable insights to optimize the services offered by specialized centers.<sup>33</sup> These data are essential to efficiently allocate resources and develop prevention and treatment strategies aligned with the needs of the patients.

## Conclusion

The present study identified the predominance of CRT and CLT, particularly among male patients, highlighting the need for early interventions and continuous follow-up, especially in more complex cases. We observed that 59.3% of the patients had already undergone surgical procedures, such as cheiloplasty and palatoplasty, indicating that many of them begin rehabilitation during the first months of life. These findings reinforce the importance of a multidisciplinary approach, which is essential to address functional and psychosocial aspects in an integrated manner, thereby promoting comprehensive recovery.

The results underscore the significance of regional epidemiological surveys to guide public policies and optimize specialized care, ultimately improving the quality of life of the patients and their families.

## Authors' Contributions

BMSOS: conceptualization, methodology, validation, formal analysis, investigation, data curation, writing – original draft, writing – review and editing, visualization, supervision, and funding acquisition; RSB: conceptualization, methodology, validation, formal analysis, investigation, data curation, writing – original draft, writing – review and editing, visualization, supervision, and funding acquisition; TAS: methodology, formal analysis, investigation, and writing – original draft; MFGB: methodology, formal analysis, investigation, and writing – original draft; JJS: methodology, investigation, data curation, and writing – original draft; CAC: conceptualization, methodology, validation, formal analysis, writing –

original draft, and writing – review and editing; MCO: conceptualization, methodology, validation, formal analysis, writing – original draft, writing – review and editing, visualization, supervision, and project administration.

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#### Clinical Trials

None.

#### Conflict of Interests

The authors have no conflict of interests to declare.

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