

UNVEILING THE COMPLEXITY: UNDERSTANDING CLEFT LIP ANOMALIES: A REVIEW

Anukriti Kumari ¹, Anurag Rawat ², Ramya Palaniappan ³, Ritik Kashwani ^{4*}, Maseer Khan ⁵ and Abdul Rahman Mohammed Al Ansari ⁶

¹ Intern, School of Dental Sciences, Sharda University, Greater Noida, India.

Email: anukritishrma0@gmail.com, ORCID ID: 0009-0001-8434-741X

² Himalayan Institute of Medical Science, Jolly Grant Dehradun, Uttarakhand India.

Email: anuragrwt@gmail.com, ORCID: 0009-0001-5534-2740

³ Madanapalle Institute of Technology and Science.

Email: ramya.btech2009@gmail.com, ORCID ID: 0000-0001-5196-0069

⁴ BDS, Ex-Junior Resident, School of Dental Sciences, Sharda University, Greater Noida.

*Corresponding Author Email: docritikkashwani@yahoo.com, ORCID ID: 0009-0008-8922-7522

⁵ Associate Professor, Epidemiology department, College of Public Health and Tropical Medicine, Jazan University, Jazan Kingdom of Saudi Arabia. Email: maseerk@jazanu.edu.sa

⁶ Salmanyia Hospital Bahrian. Email: abdulrahmanmohammedalansari@gmail.com

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Abstract

Cleft lip, a prevalent congenital anomaly affecting approximately one in every 600 newborns in the United States, presents a significant challenge in prenatal development. This multifaceted condition involves a division between the two sides of the lip, often extending to the upper jaw's bones or upper gum due to incomplete tissue fusion before birth. The complexities surrounding cleft lip encompass not only its prevalence and varied classifications but also the diverse array of challenges individuals with this condition encounter throughout their lives. Understanding cleft lip involves dissecting its prevalence, which stands as the most common birth defect in the United States, with 27,000 to 33,000 cases emerging annually in the UK. Its occurrence exhibits variations across ethnicities, with higher incidences observed in certain populations such as Caucasian and Japanese groups. Numerous factors contribute to the development of cleft lip, spanning genetic predispositions to environmental influences. Smoking, alcohol consumption, nutritional deficiencies, and medication use during pregnancy are among the environmental factors implicated in cleft formation. Genetic factors, family history, and associated syndromes further underscore the complexity of this congenital anomaly. Anatomical variations present in individuals with cleft lip depict a distinctive pattern of nasal deformities, alveolar clefting, and skeletal discrepancies, profoundly impacting appearance, speech, and feeding functionality. These complexities necessitate a comprehensive treatment approach spanning infancy to adolescence. Surgical interventions, beginning around 3 to 6 months, aim to reconstruct the lip and restore function, followed by subsequent procedures targeting speech enhancement, dental development, and skeletal corrections as individuals mature. This abstract encapsulates the multifaceted nature of cleft lip anomalies, encompassing their prevalence, etiological factors, anatomical variations, and a comprehensive treatment approach. Understanding these intricacies is pivotal in providing holistic care and support to individuals affected by cleft lip conditions throughout their lives.

Keywords: Cleft Lip, Congenital Anomaly, Prenatal Development, Tissue Fusion.

INTRODUCTION

Cleft lip stands as the most prevalent birth defect in the United States, affecting one in every 600 newborns. It signifies a division between the two sides of the lip, often encompassing the upper jaw's bones or the upper gum [1]. This occurrence transpires when the lip tissue fails to fully unite before birth. A cleft lip is a congenital deformity found in the primary palate, situated in front of the incisive foramen [2]. It can occur unilaterally, bilaterally, as a complete or incomplete cleft. Meanwhile, a cleft palate is a congenital anomaly that affects the secondary palate. It can also manifest as unilateral, bilateral, complete, or incomplete [3]. Typically, its occurrence stands at

approximately 1 in 700 births, resulting in around 1000 new cases annually in the UK. The Indian subcontinent stands as one of the planet's most densely populated areas, with India alone housing an estimated 1.1 billion people. This translates to around 24.5 million yearly births, and the incidence of clefts ranges from 27,000 to 33,000 cases per year [4]. It's more prevalent on the left side compared to bilateral clefts, with a ratio of 9:1, and it's twice as likely to occur on the left side compared to the right. Moreover, isolated cleft palate exhibits the highest incidence among females [5]. The frequency of clefting varies based on factors like characteristics, geographical location, and ethnicity. For instance, African populations show an occurrence of about 0.3 per 1000, while Caucasian and Japanese populations demonstrate rates of about 1 and 2.1 per 1000, respectively [6]. Cleft lip and palate classification encompasses several key factors. It includes the consideration of whether the cleft affects one side (unilateral) or both sides (bilateral) of the lip or palate, as well as determining if the cleft is complete, extending entirely, or incomplete, affecting only a portion [7]. Additionally, the classification involves identifying whether the individual presents with a cleft lip, cleft palate, or a combination of both conditions. Further assessment involves gauging the severity and extent of the cleft, varying from minor notches to significant gaps impacting speech and feeding [8] (Figure1). Medical professionals also factor in associated syndromes or conditions and may differentiate unilateral cases based on which side the cleft occurs. These classifications aid in tailoring specific treatment plans and interventions for individuals with cleft lip and palate, ensuring comprehensive and effective care [9]. Cleft lip and palate, while influenced by various factors, notably environmental, nutritional, medicinal, and genetic, remain significant concerns in prenatal development. Smoking and alcohol consumption during pregnancy pose substantial risks, elevating the likelihood of cleft formation [4,6]. Nutritional deficiencies, particularly Vitamin B6 and folic acid, have been linked to increased instances of clefts, with studies in the Netherlands and the Philippines highlighting this correlation. Folic acid supplementation, conversely, demonstrated a decrease in cleft incidence in North America and the USA. Zinc deficiency, critical in fetal development, was notably prevalent in mothers with cleft-affected offspring [10]. Certain medications, like corticosteroids and retinoid drugs, have been associated with cleft formation, often prescribed to pregnant women for conditions such as insomnia and anxiety. Exposure to organic chemicals and solvents also poses a risk.[11] Genetic factors, including family history, play a crucial role, with a 9% likelihood of transferring clefts from one affected parent to their child. Syndromes like Velocardiofacial syndrome are associated with cleft lip and palate occurrences, leading to what's termed as syndromic cleft palate. However, these conditions can also manifest without associated syndromes, termed non-syndromic cleft palate [12]. Further exploration into genetic factors continues to unravel complexities associated with cleft lip and palate etiology [4-6]. Patients born with cleft lips exhibit distinct anatomical differences, such as a shortened philtrum where one or both philtral columns are impacted. Anomalies in the orbicularis oris muscle cause it to attach abnormally to the cleft margin and alar wing. Moreover, these children commonly display a predictable pattern of nasal deformities: a downwardly displaced nasal septum, an anterior nasal spine of the maxilla displaced from its original position, a shortened columella, and flattened lower lateral nasal cartilage on the cleft side[13]. This often results in a widened alar base and rotated upper lateral nasal cartilage. Alveolar clefting is also frequent, potentially leading to the collapse of the maxillary arch and class III malocclusion, where upper teeth sit behind lower teeth. These

structural changes profoundly influence the appearance, speech patterns, and swallowing/feeding functionality of individuals with cleft lips. Typically, the cleft emerges at the convergence of the lateral and central segments of the upper lip, occasionally extending into the maxilla and palate [14]. Cleft lip and palate bring forth a multitude of associated challenges across various domains. Speech impediments arise due to velopharyngeal dysfunction, leading to hypernasal speech, often necessitating surgical intervention for correction [15]. Hearing problems, manifested by frequent ear infections resulting from Eustachian tube irregularities, surface within the early months of life. Dental anomalies encompass irregular tooth size, shape, positioning, and delayed eruption, affecting oral health. Feeding difficulties in infancy, hindering proper weight gain, demand alternative feeding methods. Cosmetic concerns, including difficulty in lip closure and speech production, contribute to the complexity [16]. Psychological impacts, notably depression, anxiety, and social communication challenges, form a significant aspect of the cleft lip and palate experience, affecting interpersonal interactions and self-esteem [17,18]. Patients with cleft lips exhibit distinctive anatomical variations, including a shortened philtrum, often impacting one or both philtral columns. Anomalies in the orbicularis oris muscle result in its attachment to the cleft margin and alar wing. Nasal deformities are common, marked by a downwardly displaced nasal septum, a shortened columella, flattened lower lateral nasal cartilage on the cleft side, widened alar base, and rotated upper lateral nasal cartilage [14,15]. Additionally, alveolar clefting frequently occurs, potentially causing collapse in the maxillary arch and class III malocclusion, where upper teeth sit behind lower teeth. These structural changes influence the appearance, speech patterns, and swallowing/feeding functionality in individuals with cleft lips [19]. Typically, the cleft forms at the convergence of the lateral and central segments of the upper lip, occasionally extending into the maxilla and palate [20].

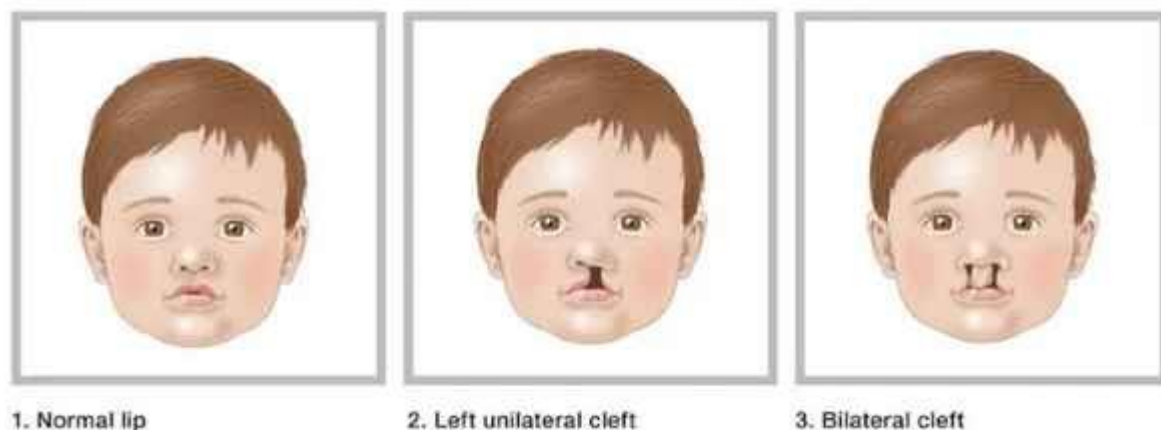


Figure 1: Cleft Lip in a Child

METHODOLOGY

The methodology for exploring the intricacies of cleft lip anomalies involves a meticulous approach using PubMed, Google Scholar, and PubMed databases. This method entails an extensive search utilizing specific keywords related to cleft lip anomalies, followed by a rigorous filtration process focusing on recent, peer-reviewed publications within the last decade. The collected data will encompass various facets, including anatomical variations, genetic underpinnings, treatment modalities,

epidemiological insights, and the multifaceted impact of cleft lip anomalies. Information will be critically analyzed, emphasizing quality and relevance, with a keen focus on ethical considerations and proper citation of sources. The synthesized findings will culminate in a comprehensive manuscript, organized systematically to provide a holistic understanding of cleft lip anomalies, subject to validation through expert review for accuracy and completeness.

DISCUSSION

Cleft lip, an intricate congenital anomaly, has been extensively examined and dissected by researchers in literature. The multifaceted discussions span across numerous facets, including etiology, epidemiology, treatment modalities, and the wide-ranging impact on affected individuals. Etiological discussions by Dixon et al. (2011) [15] and Mossey and Little (2009) [7] shed light on the intricate interplay between genetic predisposition and environmental influences in the development of cleft lip. These studies underscore the significance of comprehending the role of genetic factors alongside external influences such as maternal smoking, alcohol consumption, and nutritional deficiencies during pregnancy in the manifestation of this condition. Additionally, research by Leslie et al. (2016) [17] and Grosen et al. (2010) [18] delves into the genetic variations associated with both syndromic and non-syndromic forms of cleft lip, providing invaluable insights into the intricate genetic mechanisms underlying this anomaly. Regarding epidemiology, studies by Tanaka et al. (2015) [19] and Harville et al. (2010) [12] highlight the diverse prevalence rates of cleft lip across various geographical regions and ethnic groups. These studies emphasize the necessity for tailored preventive measures and healthcare interventions based on the regional disparities observed in occurrences of cleft lip. Treatment strategies and outcomes, as outlined by authors like Millard (1976) and Mulliken (1999), [21,22] primarily focus on the efficacy of surgical techniques in repairing cleft lips. These investigations stress the criticality of early intervention and comprehensive, multidisciplinary care in maximizing functional and aesthetic outcomes for affected individuals. Authors such as Marcusson et al. (2012) and Sitzman and Giroto (2014) [23,24] explore the psychosocial impact of cleft lip, elucidating the challenges faced by individuals in terms of self-esteem, social interactions, and psychological well-being. These discussions underscore the necessity for holistic care approaches that address not only the physical but also the emotional and social aspects of living with a cleft lip. Moreover, Metaverse, Virtual Reality and Augmented Reality also offers a transformative platform, enabling surgeons to simulate procedures, multidisciplinary teams to collaborate, and patients to access education and support, revolutionizing cleft lip care [25].

CONCLUSION

Newborns born with cleft lips face several primary concerns that necessitate attention and careful management. These challenges primarily revolve around feeding difficulties, the heightened risk of aspiration, and potential issues related to airway blockage. Managing this condition involves a sustained and comprehensive commitment to medical care, particularly focusing on addressing any accompanying congenital abnormalities and meeting the nutritional needs crucial for a healthy start in life. Surgical intervention for the initial repair of a cleft lip typically takes place between 3 to 5 months of age, guided by a set of safety criteria known as the "Rule of

10s." This rule assesses the infant's age, weight, and hemoglobin levels to ensure optimal conditions for safe surgery. Multiple well-established surgical techniques exist for repairing both unilateral and bilateral cleft lips, each tailored to the specific needs of the individual case. Following surgery, a multidisciplinary approach is vital, involving subsequent procedures at different stages of development. These procedures include cleft palate repair, alveolar bone grafting, corrections for nasal deformities, and revisions for any scarring, each performed according to the individual's developmental milestones and specific requirements.

The culmination of this care plan often involves a comprehensive evaluation around the age of 16 to 18 years. This assessment aims to determine the potential necessity for orthognathic surgery, focusing on correcting any skeletal abnormalities associated with the cleft lip deformity. This stage considers the patient's skeletal maturity and aims to ensure optimal facial and oral function as they transition into adulthood.

The treatment for cleft lip often involves a multi-staged approach spanning infancy into adolescence. Initially, the focus is on surgical repair, typically performed between 3 to 6 months of age, aiming to reconstruct the lip and restore function. This surgery not only addresses the cosmetic aspect but also plays a crucial role in improving feeding, speech, and overall facial development. The procedure may vary based on the severity and extent of the cleft, using techniques like Millard, Fisher, or Mohler repair for unilateral cases and Mulliken repair for bilateral clefts. Subsequent interventions, such as cleft palate repair around 9 to 12 months, aim to enhance speech and prevent complications related to swallowing. Alveolar bone grafting, usually performed around 7 to 9 years, closes the cleft in the gum, aiding in dental development and stabilizing the upper jaw. Throughout this process, interventions like nasal deformity corrections and orthodontic procedures are tailored to individual needs. Orthognathic surgery, typically considered around 16 to 18 years, focuses on addressing any remaining facial skeletal discrepancies for better facial balance and function, marking the comprehensive journey of cleft lip treatment.

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