Case Report

INTERNATIONAL JOURNAL OF SURGERY CASE REPORTS

OPEN

Ectrodactyly and bilateral cleft lip palate in a 2-year-old boy: a rare case report

Andi Setiawan Budihardja, PhD[a,*], Billy Martin, DDS[b], Theresia Dhearine, DDS[b], Yoanne Imanuella, DDS[b]

Introduction: Ectrodactyly (lobster claw syndrome) and bilateral cleft lip and palate are distinct congenital anomalies, yet their co-occurrence in a single individual is exceedingly rare, often indicating an underlying syndromic association such as ectrodactyly-ectodermal dysplasia-clefting syndrome. This report describes such an unusual co-presentation, complicated by a strong family history and previous unsuccessful surgical interventions.

Presentation of Case: A 2-year-old boy had congenital ectrodactyly affecting the hands and feet, and a complete bilateral cleft lip and palate. He presented with significant eating difficulties and velopharyngeal insufficiency secondary to cleft palate after two unsuccessful cleft lip and nose repairs at another hospital. His father also exhibited identical conditions, suggesting a hereditary basis. Genetic testing was not performed due to financial constraints.

Discussion: The lip-nose defects were corrected using the modified Mulliken technique with closed rhinoplasty. Six months later, primary cleft palate repair was performed via the modified Langenbeck technique, including radical muscle dissection and reconstruction of the levator veli palatini, to optimize velopharyngeal function. This case underscores the importance of thorough primary surgical repair and the potential for successful outcomes in challenging secondary repairs through advanced techniques and multidisciplinary care.

Conclusion: We described a rare and likely hereditary co-occurrence of ectrodactyly and bilateral cleft lip and palate. Despite the complexity arising from previous failed surgeries, a staged, multidisciplinary approach comprising specific reconstructive techniques yielded remarkable functional and esthetic improvements, including complete resolution of hypernasality. Thus, comprehensive, expert care plays a critical role in eliciting optimal results for complex congenital malformations.

Keywords: cleft lip, cleft palate, ectrodactyly, EEC syndrome, reconstructive surgery

Introduction

Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome is a rare genetic congenital limb malformation, with an incidence of approximately 1 in 90 000 live births; it is characterized by the absence or hypoplasia of central digits, resulting in a V-shaped cleft in the hand or foot. The other names for this syndrome names include split hand–split foot–ectodermal dysplasia–cleft syndrome, split hand, or lobster claw syndrome. This condition has been postulated to be cause by mutation in TP3 gene which is located on the long arm of chromosome 3 (3q27). It can manifest unilaterally or bilaterally and may occur as an isolated anomaly or as part of a more complex genetic syndrome. Some patients affected with EEC syndrome are noted to have chromosomal deletions or translocations on the long arm of chromosome 7[1].

Cleft lip and palate (CL/P), in contrast, is a more common congenital anomaly of the orofacial region, with an incidence of around 1 in 700 live births, resulting from the failure of fusion of the facial processes during early embryonic development[2,3]. This malformation presents with a complete separation of the lip and palate on both sides, profoundly affecting feeding, speech development, and facial esthetics, and often requiring extensive multidisciplinary surgical and rehabilitative management.

While both ectrodactyly and bilateral cleft lip and palate are congenital conditions with diverse etiologies, their simultaneous occurrence in a single individual is exceedingly rare. Such a co-presentation often prompts investigation into underlying

HIGHLIGHTS

- A 2-year-old boy presented with ectrodactyly and bilateral cleft lip and palate.
- He presented with significant eating difficulties and hypernasality.
- A large lip-nose defect persisted after previous unsuccessful cleft lip/nose repairs.
- A strong hereditary component was suspected as his father had identical conditions.
- Staged, multidisciplinary surgeries yielded good functional and esthetic outcomes.

[a]Department of Oral and Maxillofacial Surgery, Siloam Hospital Lippo Village. Faculty of Dentistry, University of Pelita Harapan, Tangerang, Indonesia and [b]Faculty of Dentistry, University of Pelita Harapan, Tangerang, Indonesia

Sponsorships or competing interests that may be relevant to content are disclosed at the end of this article.

*Corresponding author. Address: Department of Oral and Maxillofacial Surgery, Siloam Hospital Lippo Village, Faculty of Dentistry, University of Pelita Harapan, Tangerang 15810, Indonesia. Tel.: +62 21 54210130.
E-mail: abudihardja79@gmail.com; andi.budihardja@uph.edu (A. S. Budihardja).

International Journal of Surgery Case Reports (2026) 138:1588–1594

Received 23 June 2025; Accepted 4 March 2026

Published online 12 March 2026

http://dx.doi.org/10.1097/RC9.0000000000000392

syndromic associations such as ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome, or other complex genetic disorders. This case report describes the highly unusual concurrent presentation of ectrodactyly and bilateral cleft lip and palate in a 2-year-old boy. The uniqueness of this case is further underscored by identical conditions in his father, suggesting a strong hereditary component. Furthermore, this patient presented to our department for complex reconstructive surgery after two previously unsuccessful cleft lip and nose repair attempts, highlighting the persistent challenges in managing such severe and rare malformations. This report aims to detail the clinical presentation, diagnostic workup, and complex multidisciplinary reconstructive approach required for optimal care in such rare and challenging cases.

Presentation of case

This manuscript has been reported in accordance with the SCARE criteria[4,5].

A 2-year-old boy was referred to the Department of Oral and Maxillofacial Surgery for reconstruction of a severe congenital lip-nose defect. His guardians' chief complaints were significant difficulties in eating and hypernasality of speech, which had severe deleterious effects on the child's nutrition, quality of life, and development.

History and previous interventions

The patient had a birth history notable for ectrodactyly affecting his hands and feet and a complete bilateral CL/P. He had previously at the age of 1 years old undergone two attempts at cleft lip and nose repair at another clinic. Unfortunately, these prior surgical interventions were unsuccessful, leading to a significant persistent lip-nose deformity (Figs. 1 and 2) upon presentation to our department. An in-depth family history revealed that his father also had ectrodactyly and bilateral CL/P (Figs. 3–6), strongly suggesting a familial hereditary basis for these conditions. The patient or his family members did not undergo genetic testing due to financial constraints.

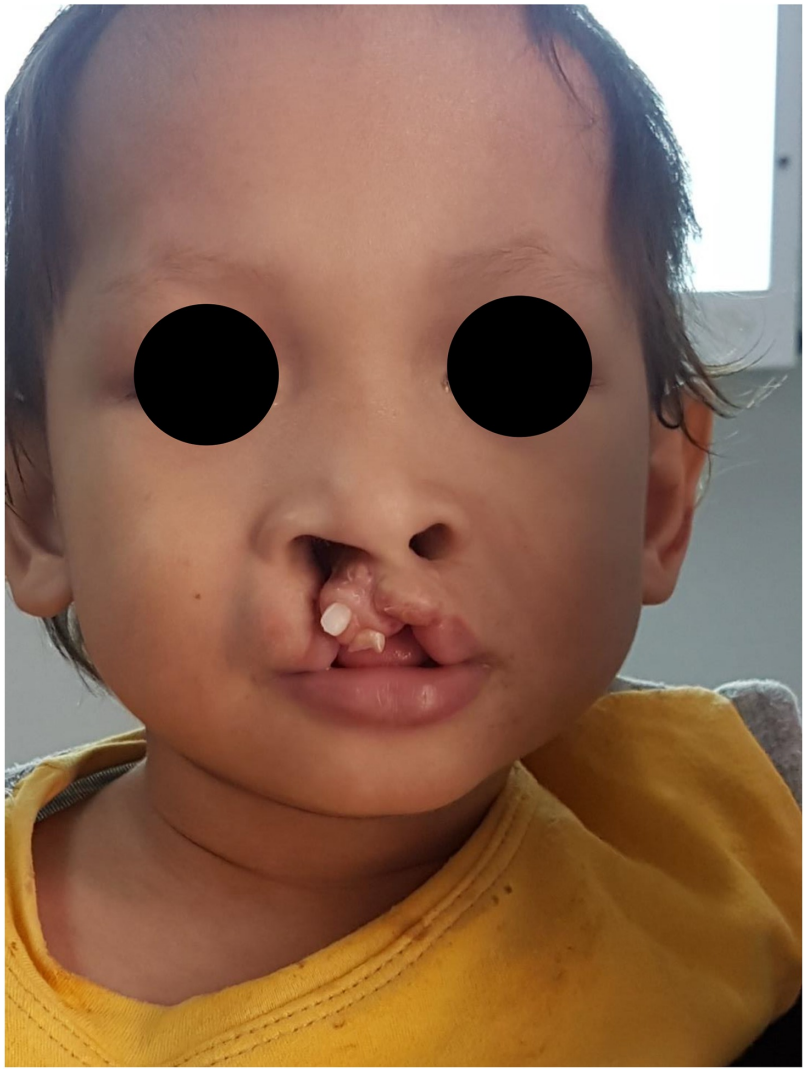

Figure 1. Lip-nose deformity after two previous unsuccessful interventions.

Figure 2. Ectrodactyly on the hand.

Clinical findings on presentation

Upon examination, the patient presented with a large, complex lip-nose defect, including a significant soft tissue defect, prominent scarring from previous surgeries, and a severely shortened columella. He also had a complete hard and soft cleft palate (Veau IV) that had not been treated surgically previously that caused severe hypernasality of the speech. Ectrodactyly was noted in both his hands and feet, characterized by typical

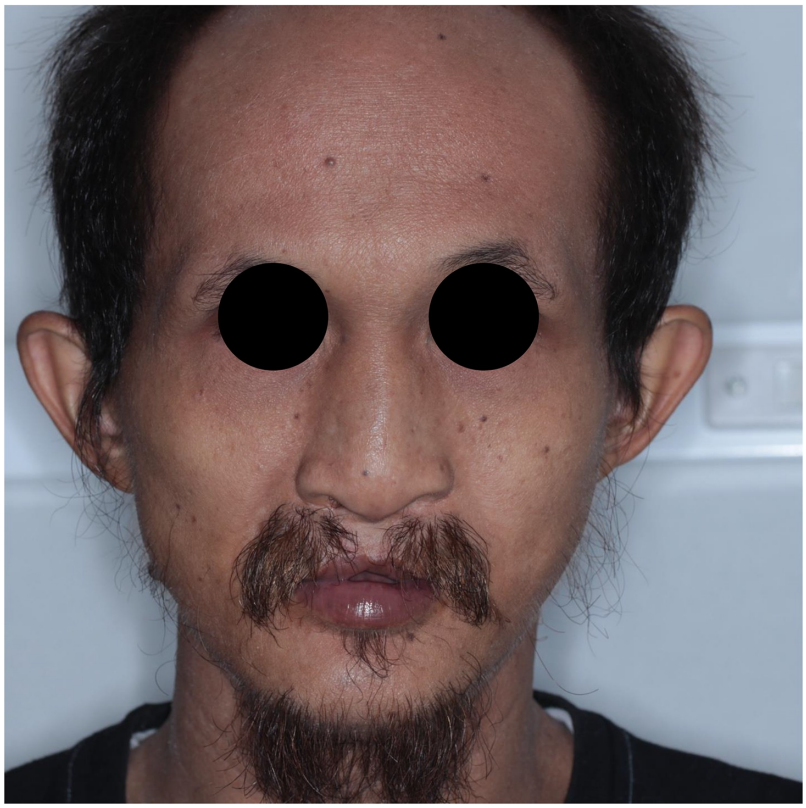

Figure 3. Father with bilateral cleft lip palate.

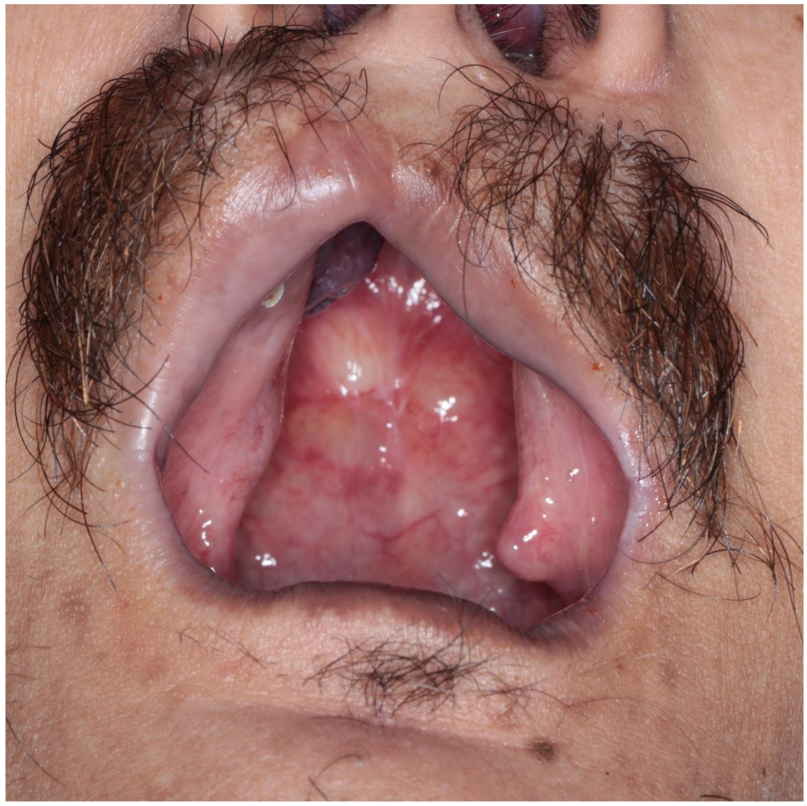

Figure 4. Complete unoperated cleft palate on his father.

V-shaped clefts and absent central digits. Besides the craniofacial and limb anomalies, other overt congenital malformations were not apparent on general physical examination.

Surgical management

Our multidisciplinary team devised a staged surgical approach. Initially, cleft lip and nose repair was performed using the modified Mulliken technique combined with closed rhinoplasty. This procedure aimed to repair the severe lip-nose deformity resulting from the previously unsuccessful surgeries. Six months later, the patient underwent primary cleft palate repair using the modified Langenbeck technique, which included radical muscle dissection and reconstruction of the levator veli palatini muscle. Radical muscle dissection was crucial to improve palatal function and ultimately correct hypernasality.

Postoperative course and outcome

After repair of the cleft palate, the patient was referred for a comprehensive speech evaluation and initiated speech therapy. At a follow-up appointment conducted 1 year after the final

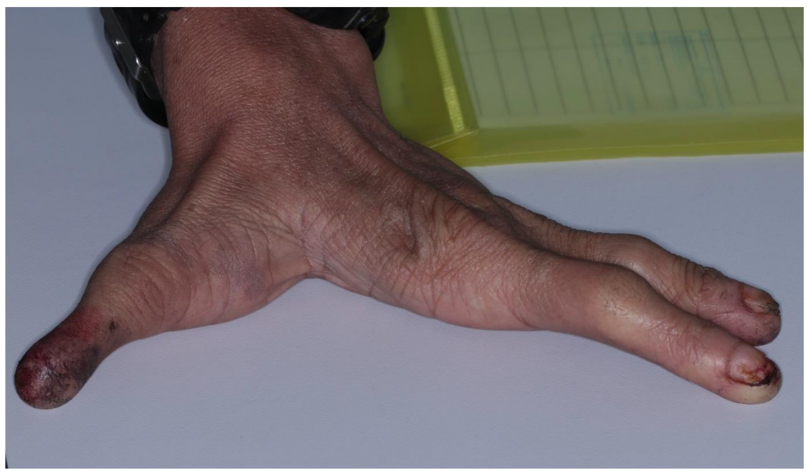

Figure 5. Ectrodactyly on the hand of the father.

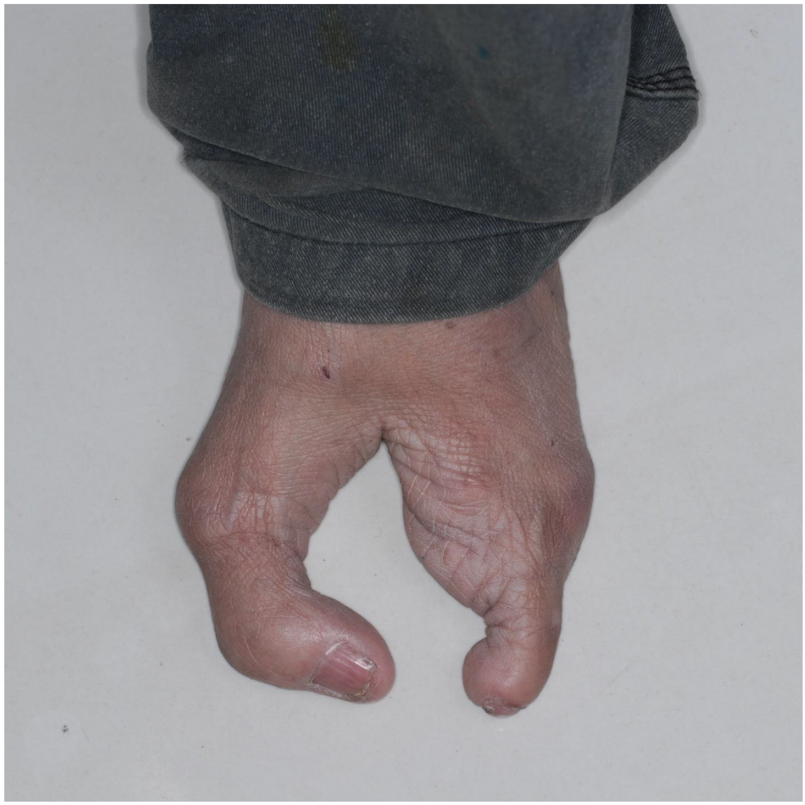

Figure 6. Ectrodactyly on the foot of the father.

surgical intervention, the patient demonstrated a remarkable improvement in both labial and nasal esthetics (Fig. 7). Crucially, his speech had significantly improved, with complete resolution of hypernasality. His eating difficulties had also resolved, contributing to better nutritional intake and overall development. The surgical outcomes were considered excellent, providing significant functional and esthetic improvement for the child.

Discussion

The co-occurrence of ectrodactyly and bilateral CL/P in a single individual, as observed in this 2-year-old boy, represents an exceedingly rare clinical presentation. To our knowledge, few such cases have been reported from Southeast Asia. While both conditions are significant congenital anomalies, their simultaneous presence strongly suggests an underlying syndromic association rather than isolated occurrences. Given the classic triad of ectrodactyly, ectodermal dysplasia, and CL/P, EEC syndrome is the primary consideration in such cases. EEC syndrome, which was first described in the literature by Rudiger[6], is characterized by the triad of ectrodactyly (hand and feet malformations) and CL/P. EEC concomitant with the tetralogy of Fallot has been reported[7]. The dermatological manifestations associated with ectodermal dysplasia include hyperkeratosis, thickened skin, and poor hair growth. Additional symptoms included dental malformation, dental caries, and abnormality of the sweat glands[8–10].

This autosomal dominant disorder is frequently linked to mutations in the TP63 gene, which plays a critical role in ectodermal development. Although EEC syndrome is usually inherited as an autosomal dominant trait, some sporadic cases can also be found[11].

The fact that the patient's father also presented with the identical combination of ectrodactyly and bilateral CL/P further supports a strong hereditary component and an autosomal dominant mode of inheritance, making genetic counseling and testing necessary in affected families. Despite its clinical importance, genetic testing could not be performed in this case due to financial constraints, which is a common challenge in resource-limited settings. Without genetic testing, parents cannot know the true reccurence risk for future children, family members who may be carriers cannot be identified, and these can cause delays in multidisciplinary treatment.

Managing such a complex case, particularly one compounded by two previously unsuccessful surgical interventions, likely due to inadequate orbicularis oris dissection, presented significant challenges. The prior surgical failures had resulted in a large lip-nose defect, necessitating a highly individualized and meticulously planned reconstructive approach. For the lip and nose repair, the modified Mulliken technique was strategically chosen[12,13] since it offers distinct advantages, particularly in the reconstruction of the central vermillion, and facilitates radical reconstruction of the orbicularis oris muscle. Meticulous muscle repair is crucial for restoring both functional competence (e.g., oral seal) and esthetic symmetry to the lip. Concomitant closed rhinoplasty was essential to address the severe nasal

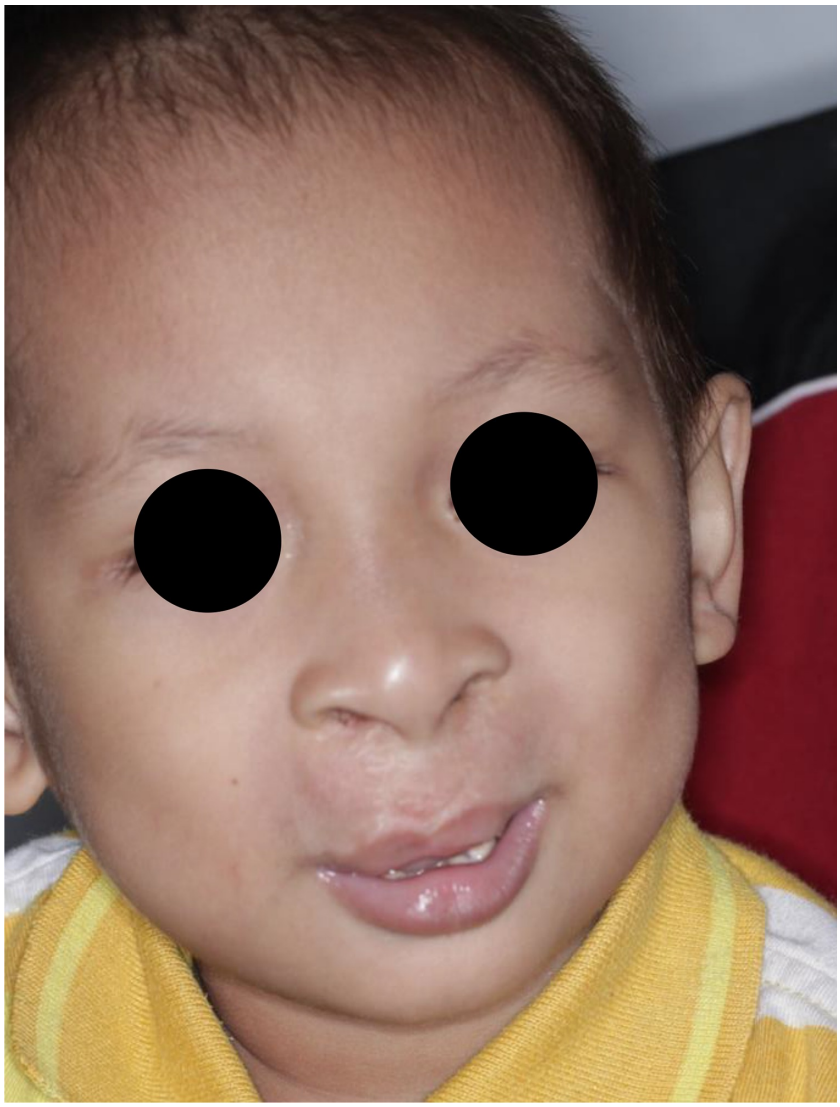

Figure 7. One-year follow-up showed good esthetic and functional outcome.

deformity often associated with bilateral clefts, which was compounded by the previous surgeries, aiming for an improved nasal form and airway.

Subsequently, the modified Langenbeck technique was employed to repair the complete hard and soft cleft palate[14]. Radical dissection and retropositioning of the levator veli palatini muscle was a critical component of this procedure. Radical muscle reconstruction is essential for achieving velopharyngeal competence, which is directly correlated with the prevention or resolution of hypernasality and the promotion of normal speech development[15]. The successful functional and esthetic outcomes, including the complete resolution of hypernasality and significant improvement in eating, underscore the efficacy of these advanced surgical techniques when applied judiciously in complex secondary repairs.

This case vividly illustrates that primary cleft lip and nose repair is of utmost importance. Well-executed primary surgery can significantly reduce the need for complex and challenging secondary reconstructive procedures, thereby minimizing patient burden and improving long-term outcomes. Furthermore, the successful management of this patient unequivocally highlights the indispensable role of a multidisciplinary approach. Comprehensive treatment includes not only advanced surgical interventions but also vigilant monitoring of nutrition, ongoing speech evaluation, and dedicated speech therapy. This holistic care model is crucial for addressing the multifaceted needs of patients with such extensive congenital anomalies and ensuring optimal functional and developmental progress.

Conclusion

This case report details the rare co-occurrence of ectrodactyly and bilateral cleft lip and palate in a 2-year-old boy with a positive family history, emphasizing the likely hereditary basis for these combined anomalies. Despite the significant challenge posed by a large lip-nose defect resulting from previous unsuccessful surgeries, a staged surgical approach employing advanced reconstructive techniques yielded remarkable functional and esthetic improvements, including complete resolution of hypernasality.

This case underscores the critical importance of meticulous primary surgical repair and the indispensable role of a comprehensive multidisciplinary team in achieving optimal outcomes for complex congenital malformations.

Ethical approval

Ethical approval for case report study is exempt/waived at our institution.

Parental consent

Written informed consent was obtained from the patient's parents/legal guardian for the publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Funding

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

Conflicts of interest disclosure

The authors have no conflict of interest to declare.

Research registration unique identifying number (UIN)

Not applicable.

Author contributions

A.S.B.: Study concept and Design data collection, Writing paper. B.M.: Writing paper, Literature study. T.D.: Writing paper. Y.I.: Writing paper and Photography.

Guarantor

Andi Setiawan Budihardja.

Provenance and peer review

Not commissioned; externally peer-reviewed.

Data availability statement

The data that support the findings of this study are available from the corresponding author upon reasonable request.

Acknowledgements

Not applicable.

References

[1] Di Iorio E, Kaye SB, Ponzin D, *et al*. Limbal stem cell deficiency and ocular phenotype in ectrodactyly-ectodermal dysplasia-clefting syndrome caused by p63 mutations. Ophthalmology 2012;119:74–83.
[2] Budihardja AS, Lutfianto B, Liman NP, *et al*. Rare facial cleft: surgical treatment and middle-term follow-up during charity operation. Craniomaxillofac Trauma Reconstr 2020;13:138–42.
[3] Budihardja AS, Nathania S. Association of obstructive sleep apnea in children with and without cleft lip palate. BMC Oral Health 2025;25:883.
[4] Kerwan A, Al-Jabir A, Mathew G, *et al*. Revised Surgical CAse REport (SCARE) guideline: an update for the age of Artificial Intelligence. Prem J Sci 2025;10:100079.
[5] Sohrabi C, Mathew G, Maria N, *et al*. The SCARE 2023 guideline: updating consensus Surgical CAse REport (SCARE) guidelines. Int J Surg 2023;109:1136–40.
[6] Rüdiger RA, Haase W, Passarge E. Association of ectrodactyly, ectodermal dysplasia, and cleft lip-palate. Am J Dis Child 1970;120:160–63.
[7] Sharma D, Kumar C, Bhalerao S, *et al*. Ectrodactyly, ectodermal dysplasia, cleft lip, and palate (EEC Syndrome) with tetralogy of fallot: a very rare combination. Front Pediatr 2015;3:51.
[8] Gupta S, Kumar S, Ray U. Lobster claw syndrome: a rare entity. Indian J Plast Surg 2013;46:158–59.
[9] Mumoli N. Lobster claw syndrome. Qjm 2012;105:1017.
[10] Sarkar S. Orodental abnormalities in lobster claw syndrome (a type of syndactyly). J Indian Soc Pedod Prev Dent 1999;17:107–10.
[11] Brunner HG, Hamel BC, Van Bokhoven H. The p63 gene in EEC and other syndromes. J Med Genet 2002;39:377–81.
[12] Allori AC, Mulliken JB. Evidence-based medicine: secondary correction of cleft lip nasal deformity. Plast Reconstr Surg 2017;140:166e–76e.
[13] Mulliken JB. The changing faces of children with cleft lip and palate. N Engl J Med 2004;351:745–47.
[14] Trier WC, Dreyer TM. Primary von Langenbeck palatoplasty with levator reconstruction: rationale and technique. Cleft Palate J 1984;21:254–62.
[15] Ghanem AM, Borg TM, Youssef G, *et al*. Surgical management of submucous cleft palate by radical muscle dissection veloplasty: speech outcomes in patients with 22q11.2 deletion syndrome. Cleft Palate Craniofac J 2024;61:498–507.