12. Rare Craniofacial Clefts

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BACKGROUND

Some 1-3% of facial clefts defy the common unilateral or bilateral pattern through the nasal floor, lip, alveolus and palate in the philtral location.1 The majority of these clefts occur sporadically, but may be present in Treacher-Collins, Goldenhar, and amniotic band syndromes. Underlying pathogenesis may be due to failure in the facial fusion process, or failure of mesodermal migration and neuroectodermal penetrance.

This chapter focuses on the principles of treating rare clefts, with detailed guidance for treating certain specific anomalies, such as midline clefts and macrostomia (Tessier 7 cleft).

KEY CONSIDERATIONS:
Rare craniofacial clefts are highly variable with respect to their severity, concomitant anomalies, and facility of repair. Some patients harboring such soft tissue clefts are otherwise completely normal, and straightforward repair can be performed in a single stage. But many have significant comorbidities or neurocognitive impairments.

Pre-surgical evaluation must address:
- Extent and degree of comorbidities
- Optimal *hospital* resources to assure safety
- Overall *treatment plan*, to achieve greatest benefit in the fewest procedures

Certain craniofacial clefts are amenable to treatment in a medical mission-type setting. Many, however, require continuity of care by a sufficiently trained and resourced team prior to embarking on surgical treatment.

RELEVANT ANATOMY

- Tessier's 1973 classification of craniofacial clefts has endured the test of time due to its clinical relevancy.
- Underlying skeletal deformities are linked to characteristic clinical findings in the soft tissue.
- Clefts are numbered from 0 to 14 and are oriented about the orbit, with clinically-observed combinations adding up to 14 (0 and 14, 1 and 13, etc)
- In 72% of facial clefts both soft tissue and bony defects exist; in 23% only soft tissues are affected, and in 5% only bone.2

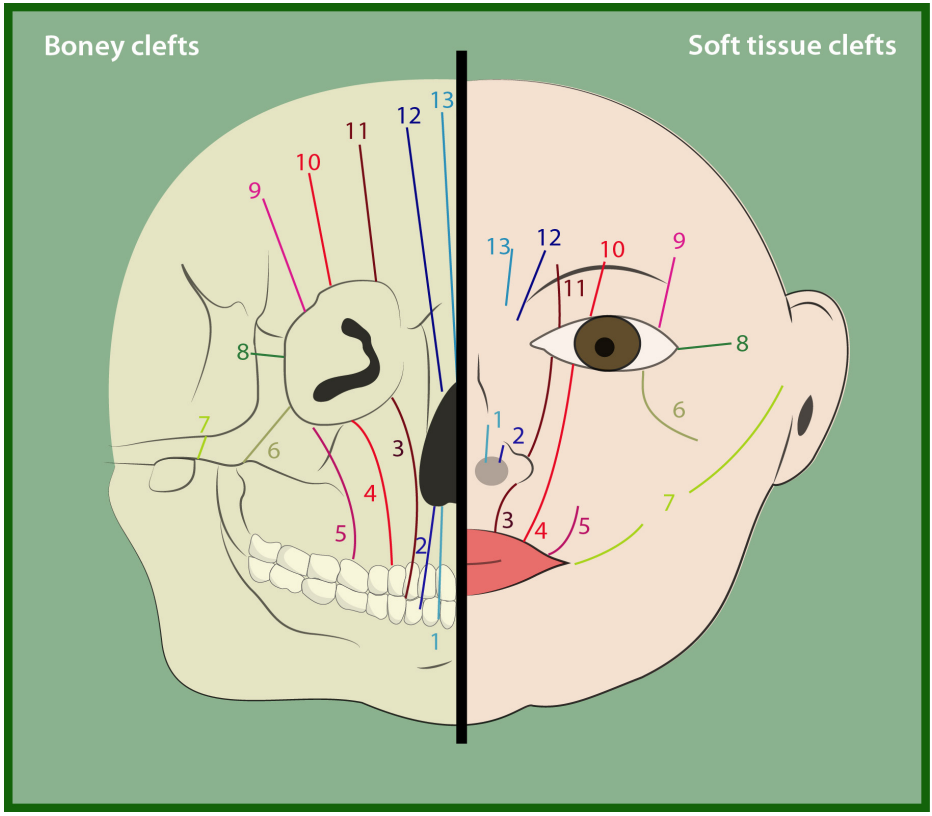
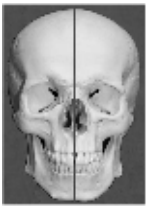
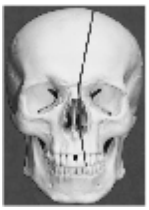
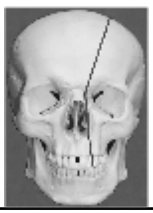
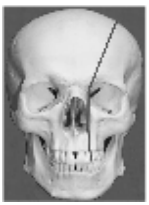
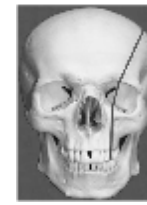
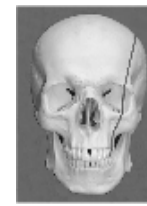
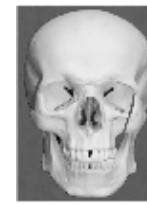
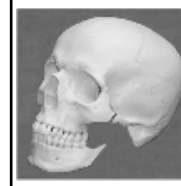
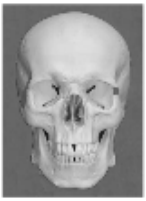
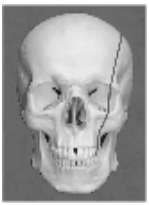
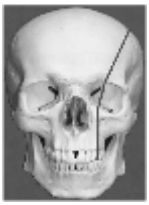
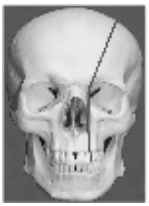
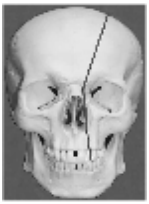


Figure 12-1. Tessier classification of craniofacial clefts of bone and soft tissue. © 2017 A Campbell, C Restrepo

Cleft	Soft-tissue characteristics	Bony characteristics	
0	With widening, a true midline cleft with central incisor diastema, bifid labial frenulum, bifid nose, duplicated septum With agenesis, a false facial cleft associated with absent philtrum, hypoplastic columella, primary and secondary cleft palate, depressed nasal tip	With widening, keel-shaped maxilla with anterior open bite, duplicated nasal spine and septum, and orbital hypertelorism With agenesis, absent nasal bone and septum, hypoplastic ethmoids, orbital hypertelorism	**0/14 Cleft** 
1	Similar to common cleft at lip Broad columella/nasal tip Cleft medial to medial canthus, causing telecanthus	Cleft between lateral and central incisors, between nasal bone and maxilla, causing hypertelorism	**1/13 Cleft** 
2	Similar to common cleft Broad columella/nasal tip Cleft medial to palpebral fissure, causing laterally displaced medial canthus	Cleft between lateral incisor and canine Passes between nasal bone and maxilla, causing hypertelorism	**2/12 Cleft** 
3	Common cleft at lip Vertical shortening between ala and lower lid, causing shortened nose and upward pull on alar base Lacrimal system disrupted Lower lid coloboma common	Cleft between lateral incisor and canine Passes lateral to nasal bone, through lacrimal groove and into orbit	**3/11 Cleft** 

4	Lateral to Cupid's bow and medial to oral commissure Ala rotated superiorly, and severe soft tissue defect between lip and eyelid Medial canthus normal	Cleft between lateral incisor and canine Travels through maxillary sinus, medial to infraorbital foramen, and through inferior orbital rim	**4/10 Cleft** 
5	Cleft just medial to oral commissure Lateral to ala and into lower eyelid	Cleft between premolars Lateral to infraorbital nerve and maxillary sinus Hypoplastic maxillary sinus	**5/9 Cleft** 
6	Similar to Treacher-Collins cleft Cheek furrowed from oral commissure to lateral eyelid Causes inferiorly displaced lateral palpebral fissure and lateral colobomas Ear/hearing defects common	No alveolar cleft Occlusal plane raised on clefted side Pierces zygomatico-maxillary suture and into lateral orbital rim Hypoplastic zygoma	**6 Cleft** 
7	Seen in facial microsomia and Goldenhar syndrome Soft tissue furrows and skin tags from oral commissure to preauricular hairline External and middle ears often malformed and occassional abnormalities of parotid gland, CN V and VII, and temporalis.	Cleft through pterygo-maxillary junction Hypoplasia of posterior maxilla and mandibular ramus Occlusal plane higher on affected side Hypoplastic zygoma and cranial base	**7 Cleft** 

8	Commonly part of Treacher-Collins syndrome Lateral palpebral fissure to temporal area True lateral coloboma and absent lateral canthus	Frontozygomatic suture cleft Hypoplastic zygoma Downward slanting lateral palpebral fissure	**8 Cleft** 
9	Extremely rare Traverses lateral third upper eyelid and brow Common CN VII palsy	Traverses superolateral orbit, greater sphenoid wing Cranial base abnormal	**5/9 Cleft** 
10	Cleft begins in middle of eyelid and brow, creating colobomas Elongation of palpebral fissure Hair tufts project from lateral brow to temple	Traverses supraorbital rim, frontal bone, orbital roof Encephaloceles and hypertelorism common	**4/10 Cleft** 
11	Cleft traverses medial eyelid and brow Downward projecting frontal hair tuft	Cleft through ethmoid air cells or lateral to ethmoids	**3/11 Cleft** 
12	Medial to medial canthus, laterally displacing canthus Possibly downward slanting paramedian frontal hairline	Traverses frontal process of maxilla, through ethmoids Produces hypertelorism and telecanthus Encephaloceles not common	**2/12 Cleft** 

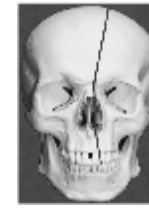
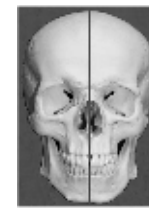
13	Medial to eyebrow, eyelids, and medial canthus Common V-shaped frontal median hair tuft	Paramedian cleft traverses frontal bone, courses along cribriform plate Hypertelorism, orbital dystopia	**1/13 Cleft** 
14	Frontonasal encephalocele Lateral displacement of orbits Hypertelorism, telecanthus	Bifid crista galli, perpendicular plate of ethmoid Short middle cranial fossa	**0/14 Cleft** 

Table 12-1. Tessier classification of craniofacial clefts. Provided by J Swanson, MD.

CLINICAL FINDINGS AND COMORBIDITIES

- Orbital *hypertelorism* is the most frequent skeletal deformation, found in 39% of craniofacial cleft patients, particularly those with 0-14, 1-13, and 2-12 clefts.2
- *Eyes* – Microphthalmia was present in 5% and anophthalmia present in 5% of patients with orbito-palpebral clefts.2
- *Encephaloceles* – Frontal encephaloceles are observed in 1% of patients. Pharyngeal encephaloceles, corresponding to a cleft of the floor of the anterior cranial fossa, are present in 0.6% of patients (each with a type 0-14 cleft; although this is only present in 2.5% of patients with a type 0-14 cleft.)2
- *Craniosynostosis* - Usually coronal suture, was found in 3% of patients and associated with multiple clefting.2
- *Cardiac Problems* – Arrhythmias are present in 3% of patients.2
- Although there are few large studies of comorbidities in patients with rare craniofacial clefts, rates of feeding difficulty, speech abnormality, and other anomalies (neurological, gastrointestinal, ocular) are likely to be similar or exceed those in patients with common cleft palate.

SURGICAL PHILOSOPHY

- Over the last 50 years, craniofacial cleft treatment philosophy has evolved.
  - Early on, when success was defined by merely closing a cleft, flap rotation and Z-plasty were used heavily.
  - With experience, high volume centers saw that the interposition of flaps from different regions of the face produced a patchwork effect because of the differences in color, texture, and thickness of the skin. Z-plasties increased the patchy effect, resulting in scars crossing the face in many directions and in conspicuous locations.[2]
  - The challenge is not closing the cleft, it is achieving a reasonable long-term aesthetic result.

- More recently, the high-volume Hospital General "Manuel Gea Gonzalez" in Mexico City has employed the following treatment philosophy:
  - Restoration of the craniofacial skeleton
  - Reconstruction with skin and soft tissue of like color and texture
  - Generous use of tissue expanders
  - Aesthetic unit and subunit reconstruction
  - Scar location at limits of aesthetic subunits
  - Symmetric repositioning of key facial landmarks

TREATMENT PRINCIPLES

- Timing of surgical intervention is guided by malformation severity.
  - If the deformity is severe or functionally encumbering (e.g. corneal exposure), early intervention is mandated; if mild, surgery can be delayed1
  - Soft tissue and cranial defects are generally corrected during infancy and prior to primary school. Midface bone grafting and correction is done after age 6. Orthognathic surgery is deferred until after skeletal maturity, age 14

Key Tip: Cheek tissue expanders have been one of the most revolutio-

nizing advancements for rare craniofacial clefts, because they enable lateral expansion and medial mobilization of tissue, which facilitates incision placement at junctions of anatomical subunits, such as the nose-cheek junction.

- Anatomic considerations:
  - *Lip reconstruction* should seek recreation of orbicularis oris continuity, alignment of the skin-vermillion junction, with incisions along the philtral column
  - *Nasal reconstruction* is facilitated by excision of redundant tissue and abnormal surrounding structures, and use of a paramedian forehead flap. Pre-expansion with a forehead tissue expander is often very helpful. Cartilage should be repaired or restored with cartilage or composite grafts.
  - *Orbital reconstruction* should utilize cranial bone grafts to restore continuity and prevent vertical orbital dystopia. If the periorbital contents have been displaced inferiorly, this can be corrected with placement of buttress grafts to the affected orbital floor.
  - *Eyelid reconstruction* can utilize transposition flaps for skin deficiency, a Fuente del Campo flap for medial canthal skin, and palatal mucosal grafting for posterior lamellar deficits. Medial canthal reconstruction should utilize transnasal wires.

SPECIFIC SURGICAL TECHNIQUES

MIDLINE CLEFTS, or Tessier number 0 clefts, are variable in severity, and some are amenable to fairly straightforward surgical correction. The following taxonomy stages midline clefts to the lip and nose.

Types of midline clefts3:
Type 1: Vermillion notch; intact orbicularis oris and pars marginalis
Type 2: Incomplete labial cleft with extension through vermillion, discontinuous orbicularis oris muscle
Type 3: Complete labial cleft; wide, divergent lower lateral cartilages
Type 4: Complete labial cleft; nasal bifidity, hypertelorism
Type 5: Wide complete nasolabial cleft, complete heminasal divergence, hypertelorism

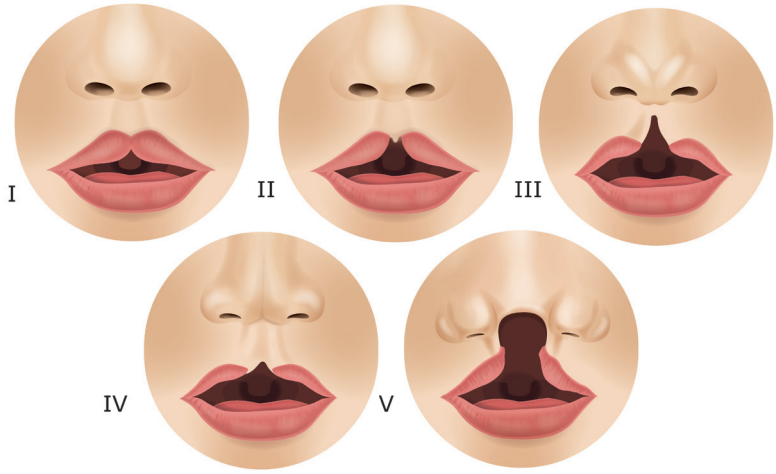


Figure 12-2. Classification of midline (Tessier 0) clefts. © 2017 A Campbell, C Restrepo

Repair techniques:

- Type 1: Vertical wedge with vertical, straight-line closure.

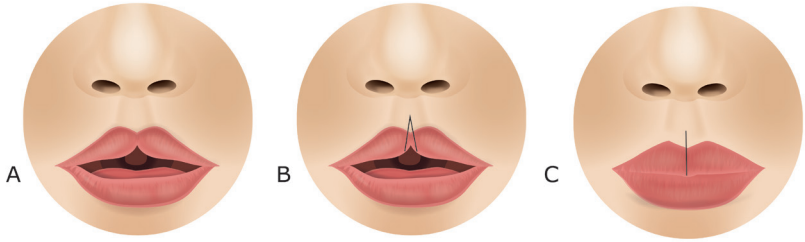
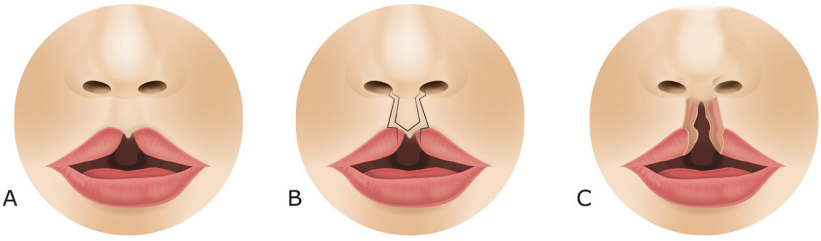


Figure 12-3. Surgical repair of Type 1 midline (Tessier 0) cleft. © 2017 A Campbell, C Restrepo

- Type 2: Wedge excision would overlengthen lip skin. Improved results with creation of a philtral shield and orbibularis muscle repair, akin to a Mulliken bilateral lip repair.



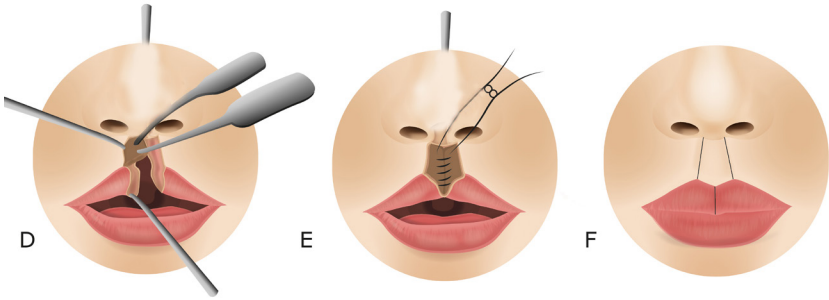


Figure 12-4. Surgical repair of Type 2 midline (Tessier 0) cleft. © 2017 A Campbell, C Restrepo

- Type 3: Elongation of foreshortened lateral lip elements with convex curvilinear full thickness skin, orbicularis, and mucosal pairings. Medial crura of lower lateral cartilages are reapproximated after redundant intervening fibrofatty tissue is excised. Z-plasty or triangular flaps can be added at the columellar-labial or skin-vermillion junctions.

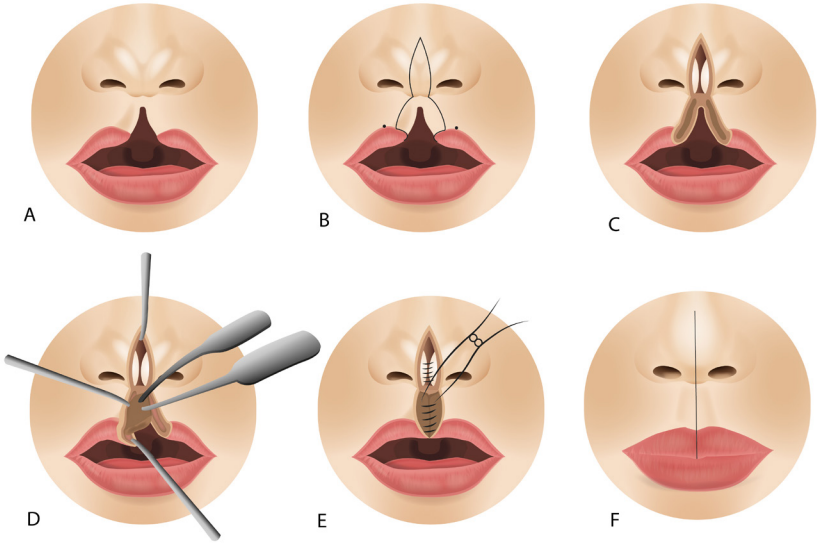


Figure 12-5. Surgical repair of Type 3 midline (Tessier 0) cleft. © 2017 A Campbell, C Restrepo

- Type 4: The treatment strategy for a Type 3 cleft is modified, with unification of the upper lateral cartilages and duplicated septa, in a fashion similar to the lower lateral cartilages. Consider a costal columellar graft in adolescence. Orbital hypertelorism is addressed

with facial bipartition, which can be performed at the same time as soft tissue repair, or orbital box osteotomy and repositioning, which is typically done at an older age.

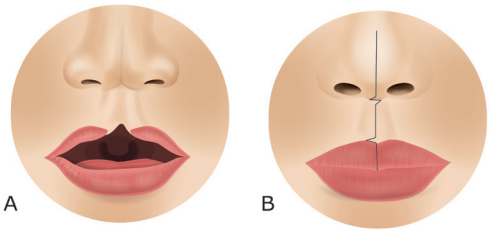


Figure 12-6. Surgical repair of Type 4 midline (Tessier 0) cleft. © 2017 A Campbell, C Restrepo

- Type 5: The treatment strategy for a Type 4 cleft is followed but with amplified naso-labial dissection and mobilization. A facial bipartition, or monobloc osteotomies, is required to restore unity of the nasal bones. A one-stage repair is advocated when possible. With maturation, midface and nasal deformity may necessitate later-staged bone graft and revision.

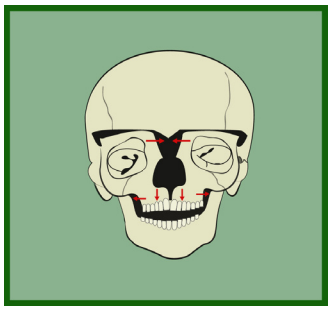


Figure 12-7. Facial bipartition for surgical repair of Type 5 midline (Tessier 0) cleft.
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- **Revision:** Particularly in secondary cases with an inadequate philtrum, reconstruction with an Abbe flap in two stages can be considered.

MACROSTOMIA is often present with **Tessier number 7 clefts**, which can be present in craniofacial microsomia (including Goldenhar syndrome), Treacher-Collins syndrome, and sporadically. The cleft can also introduce abnormalities of Orbit, Mandible, Ear, Nerve, and Soft Tissue (O.M.E.N.S. classification) and each of these should

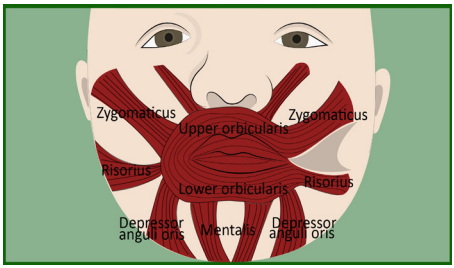

Figure 12-8. Muscular anatomy of lateral (Tessier 7) cleft. © 2017 A

be assessed.

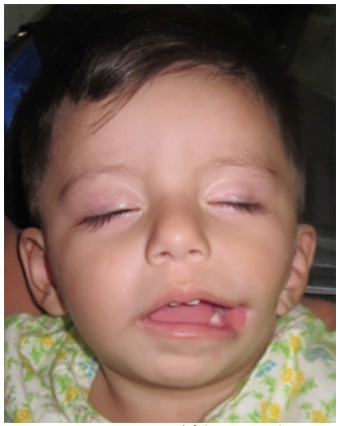

Figure 12-9. Tessier 7 cleft (macrostomia)
Source J Swanson, MD.

- Incidence ranges from 1:100 to 1:300 of all facial clefts.
- Unilateral > Bilateral
- Results from failure of the mandibular and maxillary processes to fuse during the fourth and fifth week of embryogenesis.
- May be an isolated phenomenon, but it is usually associated with other disorders such as hemifacial microsomia and / or ear tags.
- Presentation can vary from minimal displacement of the oral commissure to a complete cleft of the face.
- zTypically the overt transverse cleft terminates between the

Principles of Surgical Reconstruction:

- Repair as early as possible to assist with speech and eating (6mo.)
- Commissuroplasty is performed to close the lateral facial cleft and restore contiguity of the orbicularis oris muscle.
- Accurate positioning of the oral commissure
- Reconstruction of the orbicularis muscle is the key to functional and aesthetic construction of the commissure and is performed with an overlapping myoplasty.
- Skin closure with minimal scar
- Avoidance of late migration of oral commissure due to scar contracture
- Z-plasty to break up the scar and recruit cheek skin.
- It is generally agreed that the repair should be performed as early as possible, to prevent feeding and speech problems, as well as for aesthetic reasons.

Preoperative Markings:

- Positioning of the new commissure based on measurements from

normal side cupid's bow to the contralateral normal commissure, as well as careful inspection of the anatomic changes of the muscle and vermilion at the abnormal commissure. Normal commissure also falls along a straight line dropped from the medial limbus of the iris.

- In cases of bilateral macrostomia, the position of the new commissure is placed at the level of the medial limbus symmetric distances from midline.
- Utilizing above techniques the commissure points (c points) are marked on the upper and lower lateral lip elements at skin vermillion junction.

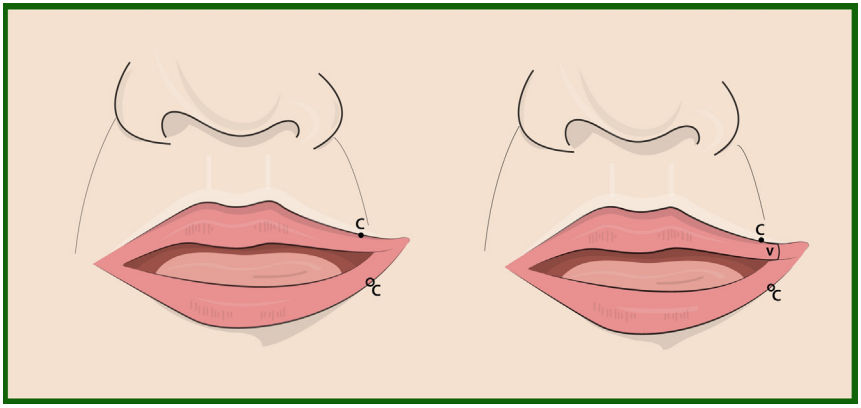

Figure 12-10. Commissure "C" points marked on upper and lower lips at skin – vermillion junction.

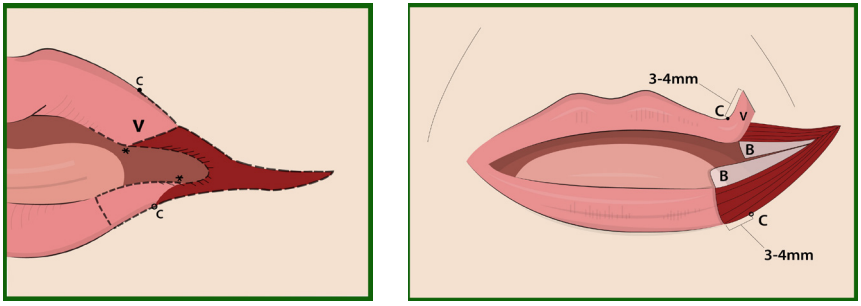

Figure 12-11. Medially based vermillion "V" flap raised and transposed across new commissure.

- A small medially based triangular vermillion flap (v flap) is raised from the upper (or lower) lip and transposed across the new com-

missure to prevent commissure migration.

- Buccal mucosa turnover flaps (b flaps) are designed on upper and lower lips to be pedicled on the internal mucosa for intraoral reconstruction.
- A Z-plasty is placed in the skin closure to break up the scar and to prevent linear contraction. The central limb of the planned Z-plasty lies along the line of the cleft. The resultant scar after transposition of the Z-plasty is best if it has central limb of the resultant scar perpendicular to the line of the cleft.

Key Tip: Incisional markings incorporate a myomucosal flap so that the incisional scar through the mucosa is moved 3-5mm away from the commissure, either onto the lower (usual) or upper lip. (This likely reduces commissure irritation, dehiscence/widening, and scar contracture at the commissure.)

Surgical Repair(4):

- Infiltration lidocaine / epinephrine
- Incisions made with 15 blade.
- V flap elevated off of musculature and based medially at upper lip c point.
- B flaps raised off of musculature and pedicled on intraoral mucosa.
- Aberrant orbicularis oris muscle dissected free from surrounding tissues laterally from cleft margin.
- The mucosa is approximated in straight-line fashion to the new commissure.
- Orbicularis muscle is repaired in an overlapping fashion with upper portion overlapping anterior to lower portion to recreate the sphincter mechanism.

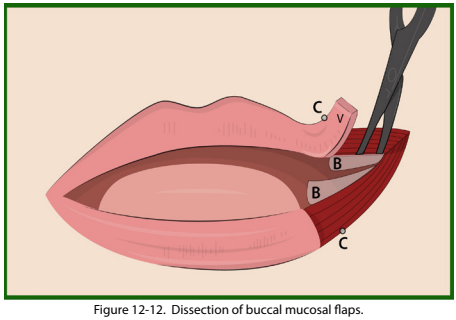

Figure 12-12. Dissection of buccal mucosal flaps.
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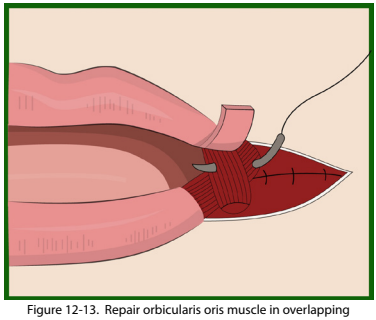

Figure 12-13. Repair orbicularis oris muscle in overlapping fashion. © 2017 A Campbell, C Restrepo

- Temporary cutaneous suture at c points can facilitate accurate reconstruction. Muscle sutures performed with prolene horizontal mattress sutures.

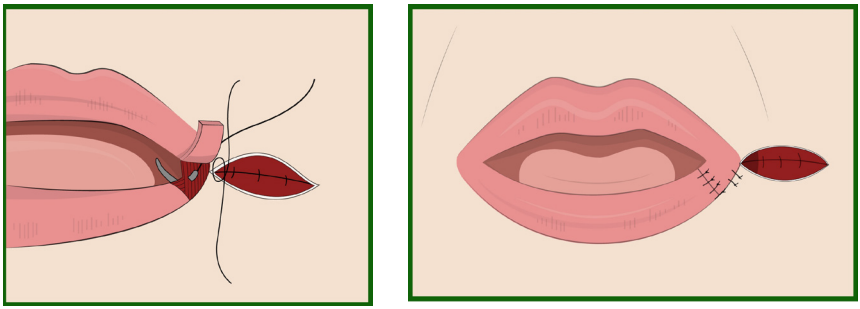

Figure 12-14. Design of z-plasty with transposition and of flaps. © 2017 A Campbell, C Restrepo

- Finally, as the skin incision is closed, a Z-plasty is placed in the incision near the nasolabial fold to lengthen the scar, break up its appearance visually, and limit contraction
- When designing a Z-plasty the medial limb should always be planned to eventually lie along RSTLs. This has been shown to result in the most aesthetically pleasing scar. It follows naturally that the lateral limb of the Z-plasty should likewise be aligned parallel to RSTLs to have equal triangular flaps in the Z-plasty to facilitate easy closure.
- Alternatives include placing multiple Z-plasties, a W-plasty, or cutting each side of the incision with intersecting triangles to create a W-plasty like incision.

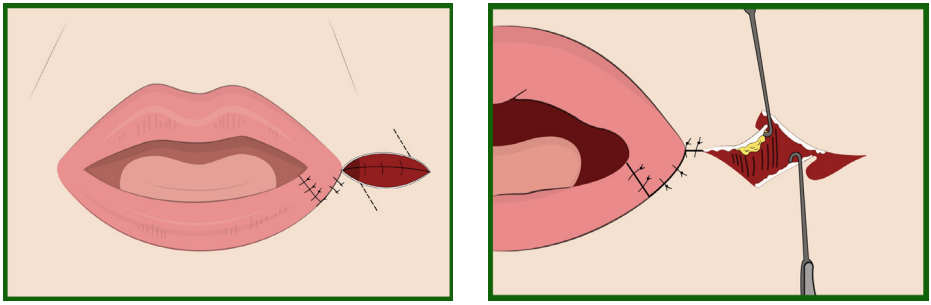

Figure 12-15. Repair of commissure with transposition and inset of "V" flap. © 2017 A Campbell, C Restrepo

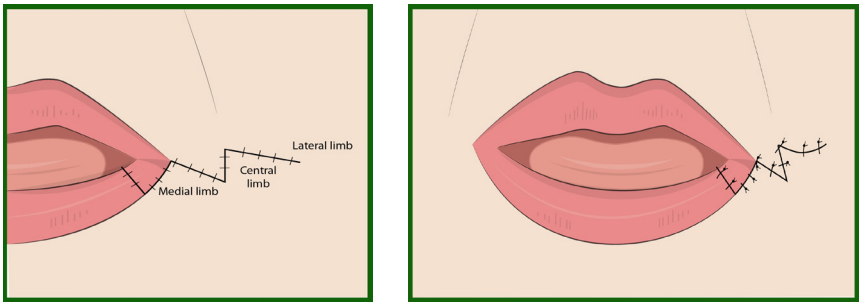

Figure 12-16. Inset of z-plasty transposition flaps and final closure of repair. © 2017 A Campbell, C Restrepo

- Postoperative care is similar to a cleft lip repair.
- Reasonable surgical treatment options exist for the majority of hemifacial deformities that frequently present with a Tessier number 7 cleft, including:
  - Soft tissue deformity: Free fat transfer, dermo-adipo-fascial graft, or free tissue transfer
  - Ear deformity/microtia: Otoplasty or ear reconstruction with costochondral graft or porex implant
  - Mandibular hypoplasia: Mandibular distraction osteogenesis, costochondral mandibular reconstruction, or orthagnathic surgical treatment
  - Nerve deficit: Contralateral neurectomy

OUTCOMES

With average 5-year follow-up in one study, patients with rare craniofacial clefts underwent an average of 3.1 operations each. This attests to the continuity of care important for the majority of these patients.2 The number of operations per patient has not decreased over time, likely because of increased emphasis on staged treatment and optimizing aesthetic outcomes.[2]

KEY READING

1. Ozaki W and Kawamoto HK. Craniofacial clefts. In Thaller SR, Bradley JP, Garri JI, eds. Craniofacial Surgery. Informa: New York, 2008.
2. Monasterio FO and Taylor JA. Major craniofacial clefts: Case series and treatment philosophy. Plast

Reconstr Surg. 2008; 122: 534-43.

3. Kolker AR, Sailon AM, Meara JG, Holmes AD. Midline cleft lip and bifid nose deformity: Description, Classification, and Treatment. J Craniofac Surg. 2015; 26: 2304-8.
4. Kaplan EN. Commissuroplasty and myoplasty for macrostomia. Ann Plast Surg. 1981; 7: 136-144.
5. McCarthy JG, Grayson BH, Hopper RA, Tepper OM. Craniofacial microsomia. In Neligan PC, ed. Plastic Surgery, 3rd Ed. New York: Elsevier. Vol 3, 761-791.
6. Bradley JP and Kawamoto HK. Craniofacial clefts. In Neligan PC, ed. Plastic Surgery, 3rd Ed. New York: Elsevier. Vol 3, 701-729.