

# Macrostomia: A Practical Guide for Plastic and Reconstructive Surgeons

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**Abstract:** Macrostomia is a rare and debilitating congenital anomaly with incompletely understood etiopathogenesis. Despite the phenotypic variability in macrostomia, plastic surgeons should demonstrate competence in the diagnosis and management of this condition. The anatomy, embryology, classification, and clinical presentation of macrostomia are reviewed in this manuscript. A historical overview of surgical repair is presented that forms the basis for understanding modern techniques of repair. Finally, an effective method of macrostomia repair is presented along with review of 5-year results. It is our intent that this guide serve as a reference for plastic and reconstructive surgeons to accomplish safe, functional, and aesthetic macrostomia reconstruction.

**Key Words:** Macrostomia, Tessier cleft number 7, transverse facial cleft

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## OVERVIEW OF MACROSTOMIA

Macrostomia is defined as an enlargement of the mouth at the oral commissure. It is associated with the Tessier cleft number 7 and is also referred to as transverse facial cleft. Macrostomia is a rare condition with a reported incidence ranging from 1/80,000 to 1/300,000 live births.<sup>1–3</sup> In contrast, the incidence of the more familiar isolated cleft lip and cleft palate is approximately 1/1000 and 0.5/1000 live births, respectively.<sup>4</sup> Unilateral macrostomia is more common than the bilateral condition; approximately 10% to 20% of cases are bilateral.<sup>5–8</sup> The left commissure is more commonly affected than the right, and men are affected more often than women. Some studies, however, demonstrate a female predilection.<sup>1,2,8</sup>

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The etiology of macrostomia remains incompletely understood. Although multiple hypotheses exist, no single explanation has emerged to explain the phenotypic variability encountered. The morbidity of macrostomia manifests as speech difficulties, oral incompetence, and difficulty with mastication and facial expression. Some or all of these may be present, in addition to the conspicuous aesthetic deformity.

Correction of macrostomia falls within the domain of plastic and reconstructive surgery. This review is designed to address the need for a current, concise, and comprehensive overview of macrostomia and provides a guide for performing an established method of repair.

## ANATOMY

The basic anatomic features of macrostomia are depicted in Fig. 1. The extent of macrostomia varies from mild lateral displacement of the commissure to a complete transverse facial cleft, including skin, muscle, and bone (Fig. 2). The direction of the cleft is horizontal or obliquely oriented toward the ipsilateral ear, and there is horizontal soft-tissue shortening on the affected side. In most patients, the defect does not extend laterally beyond the anterior border of the masseter muscle. In bilateral cases, the cleft edges on either side may be asymmetric.

An essential feature of macrostomia is the discontinuity of orbicularis oris muscle at the oral commissure. Clinical examination reveals a contour depression at the cleft bordered superiorly and inferiorly by muscular pillars that represent the terminal ends of orbicularis muscle (Fig. 1). Splaying and accentuation of the deformity occurs on animation secondary to the loss of the “check-rein” action of orbicularis and unopposed action of risorius, zygomaticus, and depressor anguli oris.<sup>9</sup>

Overlying the muscular defect at the oral commissure is a vermilion-like mucous membrane with similar histologic and clinical and properties to wet vermilion (Fig. 1). The intraoral side of the cleft is lined by normal buccal mucosa. The anatomy of macrostomia is best understood in the context and organizational topography of embryogenesis.

## EMBRYOLOGY

Development of the face takes place through differentiation and migration of the first and second branchial arches during the fifth to eighth weeks of human development. The primitive mouth or stomadeum represents a common chamber extending between the head and pericardium. The anterior opening of the stomodeum becomes bounded on its perimeter by various nests of differentiating cells or processes that ultimately form the face. These include the mandibular process inferiorly, maxillary processes superolaterally, and median nasal process and lateral nasal processes superiorly.

The orotragal line is an embryological fusion line extending from the lateral commissure to the tragus.<sup>10</sup> This line is normally obliterated with progressive fusion of maxillary and mandibular process in the fourth to fifth weeks of development.<sup>11,12</sup> This



**FIGURE 1.** Basic anatomical features of macrostomia. Anterior (left) and lateral (right) view of a child with bilateral macrostomia and soft-tissue lesions along the orotragal line. Oral commissures contain abnormal lateral extension, with thin, pale, vermilion-like mucous membrane overlying orbicularis defect. Normal buccal mucosa lines the deep aspect of the cleft.

fusion facilitates subsequent development of the muscles of mastication and facial expression, parotid gland, lateral commissure, and auricle among other structures. Failure or disruption of fusion along the orotragal line forms the anatomic basis of macrostomia.

### CLINICAL PRESENTATION

Macrostomia is often accompanied by defects of the surrounding bone, muscle, and soft tissue derived from the first and second branchial arches. Isolated macrostomia, however, is related to the first branchial arch. A recent report highlighted the variable clinical presentation of macrostomia.<sup>13</sup>

Deformities of bone accompanying macrostomia primarily involve the maxilla and mandible. The mandible may be hypoplastic, and deformities of the mandibular condyle, coronoid, and ramus are not uncommon.<sup>14</sup> Maxillary duplication with supernumerary teeth and overlap of maxillary arches has been reported in 39% of patients; up to 55% have a simple cleft of the maxillary dental arch.<sup>8,15</sup> Zygomaticotemporal cleft, absence of zygomatic arch and zygomatic bone, and deformities of the glenoid fossa and cranial base have all been reported.<sup>7,14</sup> Abnormalities of the external and middle ear may be seen in 60% to 70% of patients, and preauricular skin and cartilaginous tags are among the most common ear deformities (Fig. 1).<sup>13,16</sup> Anomalies of the parotid gland, parotid



**FIGURE 2.** Phenotypic variation seen in macrostomia. Typical unilateral macrostomia cleft (left). Unilateral macrostomia with significant skin component (right). Bilateral macrostomia with moderate lateral extension (below). Not shown is the complete transverse facial cleft, which is the most severe form of macrostomia, in which there is a full-thickness defect from the tragus to oral commissure.

duct, and trigeminal and facial nerves are more rare associations with macrostomia.<sup>5,7,10,17</sup>

Macrostomia may occur as part of a syndrome; most commonly craniofacial (hemifacial) microsomia (CFM). Craniofacial microsomia is a rare disorder involving abnormalities of the ears, facial soft tissue deficiency, nerve palsies, maxillary and mandibular hypoplasia, and pathognomonic mandibular condyle abnormalities. In addition to craniofacial anomalies, CFM may be associated with vertebral, renal, and cardiac defects. When macrostomia occurs bilaterally it is rarely associated with CFM, however, nearly 25% of unilateral macrostomia patients have CFM.<sup>13</sup> Macrostomia is also seen in Treacher Collins syndrome.

### CLASSIFICATION

Among the earliest attempts to classify orofacial clefts was by Sömmering in 1791.<sup>18</sup> In 1965, Grabb<sup>10</sup> defined macrostomia as the salient feature of his Group E classification of patients with first and second branchial arch syndrome. Tessier's<sup>14</sup> anatomic classification of craniofacial clefts was published in 1976 and remains widely used even today. In this system clefts are described by numbers 0 to 14, based on the direction of the cleft relative to the orbit.<sup>14</sup> The number 7 cleft was established as the lateral facial cleft, which centers on the zygomaticotemporal suture.<sup>14</sup> In 1983, van der Muelen et al<sup>19</sup> devised a morphogenetic classification emphasizing disrupted fusion between maxillary and mandibular processes as the basis of macrostomia. Woods et al<sup>8</sup> studied macrostomia in patients without CFM or Treacher Collins syndrome and proposed a subclassification of the Tessier system; Tessier 7a for maxillary cleft and Tessier 7b for maxillary duplication. In one of the largest series to date, Fadeyibi et al<sup>2</sup> classified macrostomia by clinical features and used this system to assess outcomes of their repair. Gleizal et al<sup>20</sup> devised a classification based on the type of repair required to address the specific deformity. Aside from Tessier's general cleft classification, no specific macrostomia classification system has garnered widespread acceptance and application.

### PREOPERATIVE ASSESSMENT

No guidelines exist for preoperative workup of patients with macrostomia. When imaging modalities are readily available, high resolution computed tomography scan of the face with three-dimensional reconstruction may define associated anomalies and assist with operative planning. Plain radiographs of the face or panoramic x-rays may be helpful when advanced imaging is unavailable. Preoperative imaging is not required and is never a substitute for detailed defect analysis and investigation for associated abnormalities by the physician.

Most advocate surgical repair as early as possible.<sup>12,21–23</sup> Habal et al<sup>24</sup> found that earlier age of surgical closure is correlated with more normal speech development. Timing is sometimes dictated by the presence of more severe deformities. More commonly, the age at medical evaluation, however, determines age at time of repair. Existing reports document repair across the spectrum of patient age ranging from the newborn period to 18 years or older.<sup>9,12,13,21</sup>

### OPERATIVE GOALS

Operative repair of macrostomia can be simplified to three goals: repair of the orbicularis oris (myoplasty), commissuroplasty, and skin and buccal mucosal closure of the excised cleft.

Orbicularis myoplasty reconstitutes the oral sphincter. As pointed out by Boo-Chai,<sup>21</sup> the free ends of the orbicularis should be brought together exactly at the commissure to prevent a “goldfish mouth” appearance, with a residual skin-vermillion web devoid of muscle at the commissure. Some authors advocate suture repair



**FIGURE 3.** Key surface anatomy of macrostomia, as pointed out by Boo-Chai in 1969. In some macrostomia defects, a triangle of skin extends onto the mucosa at the corner of the mouth (excised here), separating the 2 muscular pillars of orbicularis oris.

of orbicularis to the attached facial muscles to preserve the integrity of these connections.<sup>25</sup> Laterally or posteriorly extending clefts involve disruption of the masseter and palatopharyngeus, respectively, for which masseter myoplasty and pharyngoplasty are indicated.<sup>20</sup>

Although orbicularis myoplasty contributes to commissuroplasty, skin and mucosal repair also contribute to final commissure position, appearance, and symmetry of the mouth and lips. Meticulous attention is devoted to achieving commissural symmetry in unilateral cases, or establishing symmetric commissures in bilateral cases. One of the most important goals in macrostomia repair is prevention of postoperative commissure migration from scar contraction.<sup>17</sup>

The cheek cleft requires closure of both the buccal mucosal defect and the skin defect created in the surgical repair. Skin closure methods that minimize scarring and contracture while creating excellent contour adjacent to the commissure are central to achieving desired outcomes.

### HISTORY OF SURGICAL REPAIR

There are more described techniques for macrostomia repair than the average number of these patients a plastic surgeon will encounter in his or her professional career. No single technique is clearly superior to another, and excellent outcomes can be obtained with a variety of different techniques.



**FIGURE 4.** Setup for macrostomia repair. Endotracheal tube is oriented midline inferiorly and positioned to minimize contact with lips or face, preventing distortion of facial landmarks.



**FIGURE 5.** The border between normal and abnormal vermillion on both the upper and lower lips is marked with an obliquely oriented slant to provide an anatomic curvature to the neocommissure following cleft excision.

Among the earliest well-documented descriptions of macrostomia repair was by May<sup>26</sup> in 1962, who performed macrostomia repair using the Estlander flap.<sup>9</sup> Also in 1962, Stark and Sanders<sup>12</sup> published their account of neonatal macrostomia repair by direct excision and layered closure.

In 1969 Boo-Chai<sup>21</sup> in Singapore emphasized the importance of surface landmarks in the surgical repair of macrostomia. He noted a triangular extension of skin onto mucosa at the corner of the mouth, which was bounded by two “muscular pillars” that represented orbicularis muscle ends (Fig. 3). He recognized that the muscular pillars corresponded exactly to the transition in vermillion color from the darker vermillion of normal lip to the lighter vermillion of abnormal lip of a cleft. This color transition marks the site of the neocommissure.<sup>21</sup>

Kaplan<sup>17</sup> in 1981 proposed a method of closure involving an upper lip vermillion flap that is transposed to the lower lip to form the neocommissure, with a lower lip vermillion flap used as a buccal turnover flap to reconstitute intraoral lining. In this approach there is no suture line or scar at the commissure, which would otherwise have a propensity to contract, deform, fissure, and become painful with movement.<sup>17</sup>

Much attention has been given to the method of superficial skin and vermillion closure. Simple linear closure is advocated by some with the reported advantages of scar concealment within normal



**FIGURE 6.** The borders of abnormal cleft tissue are outlined in preparation for excision.



**FIGURE 7.** Abnormal cleft tissue is excised with a #15 blade (left). Tissue layer organization of cleft following excision (right). The skin edges are refined to create a clean skin edge oriented perpendicular to the excised wound.

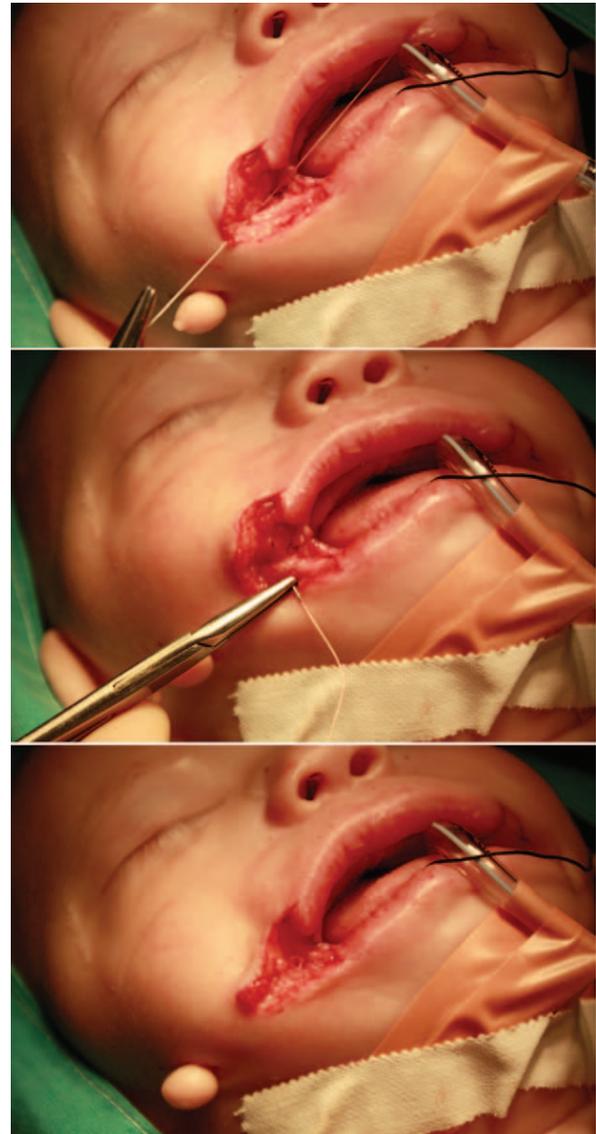
skin tension lines, natural appearance on facial animation, and superior aesthetic results.<sup>27,28</sup> Linear closure, however, has since brought about many criticisms because of increased risk of inferior and lateral commissure migration postoperatively. As a result, a host of other methods including Z plasty, W plasty, and combination techniques have since been used. Z plasty skin closure is advocated by most authors as this accomplishes lengthening of the cleft side in addition to scar disruption and stabilization of the commissure postoperatively.<sup>12,17,21,29–32</sup> In 1982 Bauer et al<sup>22</sup> published their approach using W plasty closure of the skin as an aesthetic refinement to conceal the “conspicuous cheek scar” that results from Z plasty closure.

In 1988 Verheyden<sup>33</sup> described the unique finding of a fibrous muscle band connecting the two ends of the orbicularis at the cleft, which required resection before orbicularis myoplasty. He used a mucosal-vermillion flap as the basis for neocommissure formation. Over a decade later, in 2000, Ono et al<sup>25</sup> in Japan described a method of repair involving two triangular mucosal flaps with a small Z plasty at the nasolabial fold coupled with linear closure of the skin. In this method, the orbicularis is sutured to the risorius, buccinator, or both for reinforcement following orbicularis myoplasty.<sup>25</sup> Eguchi et al<sup>34</sup> in 2001 reported the vermilion square flap method of closure, in which a lower lip pedicled mucocutaneous flap involving the vermilion border is used for neocommissure reconstruction, followed by myoplasty and “lazy W plasty” skin closure.<sup>34</sup>

In 2007 Franco et al<sup>3</sup> described a medially based myomucosal flap that is reflected medially and sutured to the opposite orbicularis after another superiorly or inferiorly based myomucosal vermilion flap is advanced for neocommissure reconstruction. The remaining portion of the cleft is then excised and closed in a linear fashion. The authors suggest that the advantage of this technique is the lack of separation of orbicularis and mucosa, preserving appropriate lip thickness and optimizing muscle function. The authors use an end-to-end muscle repair as opposed to an overlapping repair advocated by many others.<sup>17,24,28,34,35</sup> Several more recent techniques have since been reported, however they do not differ substantially from the foundations set forth by the aforementioned methods.<sup>1,36–40</sup>



**FIGURE 8.** Closure of the buccal mucosa takes place from deep to superficial, approaching the neocommissure (left). The buccal mucosa of the cleft has been closed, with remaining open skin, orbicularis, and vermilion components (right).



**FIGURE 9.** Orbicularis myoplasty. Orbicularis myoplasty begins laterally with progressive medial advancement with interrupted sutures (upper). Advancement myoplasty continues with interrupted sutures toward the neocommissure (middle). Myoplasty completed to the level of the neocommissure (below).

### SELECTED REPAIR TECHNIQUE

A 4 month old otherwise healthy girl presented for evaluation (Fig. 1). Once preoperative workup revealed no additional comorbidities, she was taken to the operating room for bilateral repair of macrostomia and excision of soft tissue facial lesions.

### Setup and Marking

Regardless of the setting in which repair occurs, preoperative setup and the markings are the most important aspects of the operation. The patient is positioned supine on the operating table with the face directed forward and midline. Ensure that the neck is neither flexor nor extended beyond the neutral straight position. For adolescent and adult patients with sufficient understanding and good health status, consideration should be given to repair under



**FIGURE 10.** Completion of myoplasty medially establishes definitive neocommissure position and aligns vermilion and skin layers for final closure.

local anesthesia, which has documented success in cleft lip repair.<sup>41</sup>

For both unilateral and bilateral macrostomia cases, the endotracheal tube is directed inferiorly in the midline. The tube is positioned to exit the mouth without contacting the lips, and tape is used to create a mesentery around the tube before it is gently secured to the chin under no tension and without facial distortion (Fig. 4). A saline moistened throat pack is placed in the posterior pharynx and affixed with a suture tag to ensure safe removal at the end of the operation. A strip of tape is used to keep the eyes closed and the face is then prepped with antiseptic solution.

Markings begin with normal anatomic landmarks; the contralateral normal oral commissure is marked (if present), along with the midline of the upper and lower lips at the vermilion border. The Cupid's bow and philtral columns are outlined. A useful landmark for neocommissure positioning is the ipsilateral pupil, which can be marked with a vertical line on the cheek. The most crucial markings define the superior and inferior borders of the vermilion at the neocommissure (Fig. 5). The borders of the abnormal mucosa and skin are outlined and the position of the neocommissure established and marked (Fig. 6).

### Operative Execution

Once the position of the neocommissure is established, a 15 blade is used to excise the marked abnormal skin, vermilion, and mucosa down to the level of the superficial subcutaneous tissue (Fig. 7). Orbicularis muscle is identified, and minimal undermining is performed of both the skin and mucosa on either side of the orbicularis.

The orbicularis muscle of both upper and lower lips has been freed from external skin and buccal mucosa. The buccal mucosa is then closed linearly from deep to superficial and ceases once the



**FIGURE 11.** Closure of remaining skin and subcutaneous tissue defect. Intermittent Z-plasty closure of skin is performed along the length of the incision.



**FIGURE 12.** Immediate postoperative repair (upper). A 2 year postop frontal views at rest and during mouth opening (center left and right). There appears to be mild lower lip excess and migration of the left commissure laterally, resulting in resting lip misalignment. A 5-year postoperative frontal view demonstrating excellent commissure position bilaterally with significantly improved resting lip alignment (below left and right).

level of the neocommissure is reached (Fig. 8). This is the first component of commissuroplasty. The two ends of the orbicularis muscle lay in close proximity laterally, with increasing distance apart medially. Progressive suture closure of orbicularis then proceeds in a lateral to medial direction, in a manner analogous to closing up a zipper on a pair of jeans (Fig. 9). With each interrupted orbicularis suture, the depth and level of purchase of orbicularis on each side must be equivalent. The final medial suture of the orbicularis myoplasty establishes the neocommissure and must be repeated till the neocommissure is perfectly aligned (Fig. 10). Orbicularis myoplasty juxtaposes the vermilion and mucosal layers of the upper and lower lips, permitting completion of commissuroplasty.

Following commissuroplasty, there remains a lateral soft tissue defect of skin and subcutaneous tissue. This is closed in a layered fashion, first with buried dermal sutures followed by skin sutures. The underlying neurovasculature is protected by avoiding placement of excessively deep sutures. Z-plasty flaps are developed along the length of the incision to break up the straight-line scar (Fig. 11). This patient was seen at 2 and 5 years postoperatively (Fig. 12).

### COMPLICATIONS

The most important complications following macrostomia repair relate to scar formation. Hypertrophic, contracted, and hyper- or hypopigmented scars may result in suboptimal aesthetic and functional outcomes. Scar contracture is associated with commissure migration that may compromise oral competence and may require operative correction. Conversely, scar contracture may also prevent complete opening of the stoma, which may also require intervention. Inadequate orbicularis myoplasty may result in an overly lax and long

lower lip, creating a “goldfish mouth” appearance.<sup>21</sup> Wound dehiscence, bleeding, and postoperative infection may also occur, however the propensity for these outcomes is not well documented.

## CONCLUSIONS

Macrostomia is a rare and debilitating congenital defect with incompletely understood etiopathogenesis. A review of macrostomia has been performed along with description of a reliable and effective method for attaining the stated goals of repair. It is our intention that this review assists plastic and reconstructive surgeons in the understanding and management of macrostomia regardless of the degree of prior familiarity with the condition.

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