Paparella: Volume III: Head and Neck

Section 2: Disorders of the Head and Neck

Part 2: The Oral Cavity

Chapter 12: Congenital Defects of the Oral Cavity

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There are numerous congenital defects of the oral cavity and its neighboring structures. Some are gross and relatively common, such as cleft lip and palate, whereas others are minor and rare, such as lateral fistulas of the soft palate. Not all of these deformities can be explained by our present knowledge of embryology. On the contrary, some of our accepted concepts of embryonic development may readily be challenged on the basis of some of these congenital deformities.

Because clefts of the lip and palate constitute the most common of the gross congenital deformities of the oral cavity, only they will be described in some detail. However, some of the less common deformities need to be mentioned for the sake of completeness, and only those of particular interest to the otolaryngologist will be described.

The following is a classification, by no means complete, of various congenital oral lesions according to their anatomic distribution:

I. Lips
   - A. Clefts
     1. Median.
     2. Paramedian. These oronasal clefts are the most common variety of lip clefts and are described at the end of this chapter.
     3. Oblique (oro-auricular).
     4. Transverse. Oroaural clefts, or macrostomia, may present on one or both sides and may vary in their extent; they occur along the line of fusion of the maxillary and mandibular processes of the face.
   - B. Microstomia.
   - C. Pits. These mucosa-lined sinus tracts usually present as symmetric pits in the vermilion border of the lower lip and are invariably associated with maxillofacial clefts. They extend vertically down the substance of the lip for a depth of up to 2.5 cm and communicate with the underlying minor salivary glands. They may, therefore, transmit a mucoid secretion to the surface. When situated anteriorly, the vermilion-cutaneous border may bulge forward in relation to the openings of the pits. They are usually excised for cosmetic reasons. Similar pits may also be present in the vermilion at the oral commissure of patients with cleft lips and on the mucosal surface of the upper lip in the midline or close to it.

D. "Double lip". This condition is produced by an apparent horizontal duplication of the vermilion of the upper lip and can be seen only when the lips are parted, especially in a smile. Actually, it is caused by a redundant mucous lining of the lip, and this redundancy may be excised for aesthetic reasons. At times this condition may be associated with blepharochalasis and a nontoxic goiter (Ascher's syndrome).

E. Neuromuscular agenesis.

II. Labial Sulci

A. Lacking. The maxillary labial sulcus is frequently lacking or very shallow in bilateral clefts of the lip. This may also be encountered in chondroectodermal dysplasia.

B. Hyperplastic frenula. These are invariably associated with a diastema of the teeth at the site of origin of the frenulum. Prior to undertaking orthodontic closure of such a diastema, the attachment of the frenulum at the crest of the alveolar process should be resected.

C. Multiple frenula. These are associated with chondroectodermal dysplasia.

III. Dental

A. Teeth.

1. Agenesis, partial or complete.
2. Supernumerary. Refers to an additional tooth, or teeth, in the series.
3. Malformations.
    a. Peg incisors.
    b. Gemination. Because of the splitting of a single tooth germ, the crown of the tooth appears bifid. There is usually a single root.
    c. Fusion. This results from the union of two tooth germs, and the two teeth may be united along their entire length or joined only at their crowns or their roots.
    d. Odontoma.
4. Syphilitic teeth.
    a. Notched incisors (Hutchinson's teeth).
    b. Domed molars (Moon's teeth).
    c. Mulberry molars.

B. Enamel.

1. Staining or mottling. This may be due to the ingestion of tetracycline or of fluorides in excess of one part/million during the tooth development period.
2. Dysplasia.
    a. Hypoplasia. Known as amelogenesis imperfecta when hereditary.
    b. Hypocalcification.

C. Gingivae.

1. Macrogingivae. This is a form of idiopathic gingival fibromatosis, which may be associated with hypertrichosis.

IV. Alveolar process
- A. Hypoplasia.
- B. Hyperplasia.
- C. Clefts.
  1. Maxillary: lateral or median.
  2. Mandibular: median.

V. Jaws
- A. Micrognathia or brachygnathia.
- B. Dysostosis (mandibulofacial).
- C. Hemihypertrophy. This condition, sometimes called Curtius' or Steiner's syndrome, may involve part or all of the face and may also be accompanied by enlargement of half of the body on the same side.
- D. Cysts.
  1. Maxillary.
     a. Midline.
        (1) Median alveolar.
        (2) Nasopalatal.
        (3) Median palatal.
     b. Lateral.
        (1) Globulomaxillary.
        (2) Nasolabial.
  2. Mandibular, median.

VI. Tongue
- A. Aglossia.
- B. Microglossia.
- C. Macroglossia. May be idiopathic or due to lymphangioma or hemangioma; also a fairly common feature of mongolism.
- D. Bifid.
- E. Ankyloglossia.
  1. Maxillary. In very rare cases the tongue may be ankylosed to the hard palate or to the alveolar ridge. It may even be attached to the lower edge of the septum if the condition coexists with a cleft palate.
  2. Mandibular. This is due to varying degrees of underdevelopment of the lingual frenulum. If it interferes with lingual function, as in speech, it is best treated by a Z-plasty if the restraining web is broad. Fine webs may be treated successfully by snipping with scissors.
- F. Lingual thyroid.
- G. Fissured (scrotal) tongue. This is rarely congenital but may appear in childhood.
- H. Median rhomboid "glossitis". Because of faulty development, the tuberculum impar appears on the surface of the tongue in front of the foramen cecum as a rhomboid wedge between the lateral halves of the tongue. It is devoid of papillae and appears red; hence, the erroneous use of the term *glossitis.*
- I. Hypertrophic papillae. A common finding in mongolism.

J. Inclusion dermoid. Usually found anteriorly in the midline and may reach large proportions.
K. Epidermolysis bullosa (dystrophic type). This rare condition is characterized by bullous and vesicular eruptions of the skin and mucous membranes and is manifested at or shortly after birth. The bullae usually heal by hypertrophic scars, leading to marked deformity of the oral cavity.

VII. Bony Palate
- A. Clefts. Palatal clefts are described at the end of this chapter.
- B. High palatal vault. May be found in conjunction with micrognathia and glossoptosis (Robin's syndrome) and may present as a respiratory emergency of the newborn. Not infrequently, it may also be encountered in Turner's syndrome and in mongolism.
- C. Cysts.
  1. Median.
  2. Incisive canal.
  3. Globulomaxillary.

VIII. Soft palate
- A. Clefts.
- B. Hypoplasia. Usually associated with submucous cleft palate (see further on).
- C. Neuromuscular agenesis. Both hypoplasia and neuromuscular agenesis may produce velopharyngeal incompetence. This may manifest itself early in life by nasal regurgitation of food.
- D. Lateral fistulas. Sometimes associated with congenital lack of the tonsils.

IX. Mucous Membranes and Connective Tissues
- A. Syphilitic stigmas.
- B. Encephalofacial angiomatosis (Sturge-Wever syndrome).
- C. Hemangioma.
  1. Capillary.
  2. Cavernous.
- D. Lymphangioma and hygroma.

Maxillofacial Clefts

One of the most frequent of the serious congenital malformations, cleft lip or palate, or both, occurs approximately once in every 700 births. The incidence rises sharply when there is a familial history of a similar deformity. It is fascinating that cleft lip and palate are more common in male infants, but cleft palate by itself is more frequently found in female infants. Clefts are far more common on the left side, a factor unexplainable on the basis of our present knowledge of embryology.

Embryologic Considerations

Some knowledge of the relevant embryologic background will aid in the evaluation and understanding of the problem. In a normally developing fetus there is fusion of the median nasal process with the maxillary process to form the upper lip, the premaxilla, and

the corresponding segments of the alveolar process - the premaxilla and its alveolus forming the primary palate. The palatal processes fuse with each other, forming the secondary palate, as well as with the nasal septum and the premaxilla between the eighth and tenth weeks of gestation. This fusion starts in the vicinity of the future incisive foramen, and from there it proceeds posteriorly and anteriorly. In cleft cases, only a tenuous epithelial fusion is said to take place, but this breaks down because of the failure of ingrowth of mesodermal tissue. Cleft palate, therefore, results from a lack of cohesion of the palatal segments and may be seen in conjunction with clefts of the lip and alveolar process or alone. Because embryonic fusion of the constituent segments is initiated at the incisive foramen, incomplete lip-palate clefts may be present only at the anterior or posterior edges of these structures, or at both edges; however, they very rarely appear as fistulas in the intervening areas, and deep midline pits also may be encountered occasionally.

Varieties of Clefts

Maxillofacial clefts have been classified in various ways.

Clefts of the Lip

The simplest kind of cleft lip is that which does not have an underlying cleft of the premaxilla on the same side, although a notch may be present in the corresponding alveolar process. Such clefts may be partial or complete, unilateral or bilateral. Incomplete clefts of the lip may extend in varying degrees from the vermilion border to just short of the floor of the nasal vestibule. Complete clefts extend into the floor of the naris, but these clefts rarely occur without some degree of involvement of the underlying premaxilla. In a unilateral cleft involving only the lip and its corresponding alveolar process, the median or premaxillary segment usually projects labially - apparently deflected in the direction of the muscle pull from the larger noncleft portion of the upper lip.

An overt nasal deformity accompanies most cleft lips, although this may not be readily apparent in some partial clefts nor obvious to the inexperienced observer. Varying in degree with the severity of the cleft of the lip, the nasal defect usually consists of a depression in the lateral crus, which may also involve the dome, deflection of the tip toward the cleft, a shorter columellar border, a wider vestibular floor, and diminished size of the ala. The membranous triangle, or facet, of the nasal lobule is often diminished or lacking on the cleft side owing to downward displacement of the dome of the tip cartilage.

When a complete cleft is present on both sides, the lip malformation is usually associated with a complete bilateral cleft of the premaxilla and invariably of the entire palate. In such a case, the premaxillary lip segment, or prolabium, is completely unrestrained by any attachment to either segment of the maxilla and is attached to the tip of the nose by a very short or almost nonexistent columella. There is usually almost a complete lack of any labial sulcus under the prolabium. Accompanying the bilateral cleft lip is a wide cartilaginous nasal vault with flared alae that are stretched out across the maxillary cleft on each side.

Complete Unilateral Cleft Palate and Lip

Complete clefts of the palate are termed *unilateral* when the palatal process of the opposite side is fused with the nasal septum. In these cases, the derangement in the palatal arch is characterized by the total lack of continuity of structure across the midline of the palate on the side on the cleft, and the premaxilla is separated from the alveolar process on the involved side. The vomer characteristically is horizontally inclined and is attached to the palatal process on the normal side so that the oral cavity communicates only with the nasal chamber of the cleft side. Often the free edge of the premaxillary segment protrudes into the cleft of the lip and may also intrude into the void in the floor of the nasal vestibule. Occasionally, the premaxillary segment may appear to overlap the free edge of the alveolar process of the maxillary segment, when the latter has rotated medially - a phenomenon that frequently follows repair of the lip.

Bilateral Cleft Palate and Lip

The frequency of bilateral clefts has been variously reported as 9.6 to 16 per cent of the entire maxillofacial cleft series. These infants have the greatest deformity of all. The cleft through the secondary palate is in the midline and divides at the incisive foramen to separate the premaxilla from the rest of the palate. The small anterior segment of the palate thus remains suspended from the anterior portion of the nasal septum. Extending backward from the premaxilla, the inferior margin of the vomer is free and appears to be thickened but is vertical and in the midline. A bilateral cleft of the palate may be termed *incomplete* when the vomer is only partially attached to one of the palatine shelves.

The incidence of a protrusive premaxilla in bilateral cleft palate is high, and not infrequently the palatal segments appear small and retruded in relation to the premaxilla. There is often disproportion between the size of the premaxilla and the constituents of the secondary palate.

In some cases, there may be adequate space between the anterior edges of the palate to accommodate the premaxilla, whereas in others, the palatal segments may have collapsed immediately behind the primary palate, leaving no room for it in the alveolar arch.

Clefts of the Palate Only

Clefts that are limited to the secondary palate and that do not involve the alveolar process are a group by themselves, both genetically and morphologically. They are more frequent in females. It is this type of cleft palate that exhibits a higher incidence of mandibular micrognathia with glossoptosis, the so-called Robin syndrome.

Midline palatal clefts may vary in extent from a submucous cleft through a bifid uvula to a complete cleft of the secondary palate as far forward as the region of the incisive foramen.

A submucous cleft of the palate refers to a condition of midline mesodermal deficiency between the nasal and oral mucous membranes involving the velum and the posterior edge of the hard palate; the latter is manifested by varying degrees of notching at

the point at which the posterior nasal spine would normally be present. Submucous palatal clefts invariably have accompanying deficiencies of the musculature of the soft palate, as may be evidenced by a short palatal velum. This condition frequently accompanies a bifid uvula. In fact, it has been this writer's experience that nearly every instance of a cleft of the soft palate has been associated with a submucous cleft of the hard palate of varying degree.

In addition to the clefts discussed, there are median maxillary clefts, clefts of the mandibular process, and the oblique facial clefts involving the ocular and aural apertures. As these clefts are rather rare, they will not be described here.