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Cleft Lip and Palate

Cleft lip and palate assume many forms. These structural defects occur very early in the development of the baby-to-be. A cleft of the lip or palate can occur separately, although these defects are more likely to occur together. Clefts can occur as isolated defects, but more often they occur with at least one minor or one major associated malformation. Approximately 70% of children with cleft lip with or without cleft palate (CL ± P) and 50% of children with cleft palate only (CPO) have no other abnormal findings (Calzolari et al., 2007; Jones, 1988). This means that individuals with CPO are much more likely to have associated anomalies. Male infants are more vulnerable to CL ± P than to CPO, and female infants are more vulnerable to CPO than to CL ± P.

Please see pages 24 to 27 in Peterson-Falzone et al. (2010) for references on these points and information regarding frequency of occurrence by type of cleft and gender, the frequency of occurrence by racial groups, and recurrence risks.

TYPES OF CLEFTS

CL ± P may be either unilateral or bilateral and incomplete or complete. The latter designation speaks only to whether any tissue is present across the line of the cleft, although the amount of tissue may be minimal. It helps to keep in mind that, in addition to unilateral versus bilateral, clefts vary in three other dimensions: anterior to posterior, width, and vertical depth or top-to-bottom (nasal mucosa, bone of the hard palate or muscle of the soft palate, oral mucosa). Even the dichotomy of unilateral versus bilateral is not always helpful in describing severity because some unilateral clefts are wider than some bilateral clefts. Thus a clinician who has not seen the child from early infancy will not be aware of the severity of the original defect and the influence of that severity on both the success of surgical repair and the development of speech.

Median clefts, oblique facial clefts, and lateral facial clefts are all very rare forms of clefts and are not discussed here. See pages 19 to 20 of Peterson-Falzone et al. (2010) for illustrations.

Although the terminology may seem confusing without taking a course in embryology, the ability to conceptualize types of clefts will be enhanced if it is noted that the middle portion of the lip (that portion extending from one lateral incisor to the other lateral incisor) and the anterior portion of the hard palate are formed from an embryonic structure termed the *primary palate*. This structure is triangular, with the tip of the triangle corresponding to the incisive foramen. The remainder of the hard palate and all of the soft palate form from the embryonic *secondary palate*.

Some clinicians term a cleft of the lip or a cleft of the lip and alveolus a *cleft of the primary palate*. Similarly, a cleft of the palate only may be termed a *cleft of the secondary palate*. Unfortunately, too many clinicians mistakenly think that "primary palate" means hard palate and "secondary palate" means soft palate.

In the embryo, the development of the primary palate begins in the region of the incisive foramen and *moves forward*. Thus the last part to form is the lip itself. The development of the secondary palate is just the reverse: It starts with the region of the incisive foramen and *moves posteriorly*. The last part to form is the uvula. A cleft of the lip and palate is thus the result of two problems in embryonic development, one in the primary palate and one in the secondary palate, either of which may be incomplete or complete.

CLEFT LIP

Clefts of the lip vary from a small defect (Fig. 1-1, *A*) to a complete cleft extending through the floor of the nostril (Fig. 1-1, *B*). Microforms of cleft lip may include a minimal notch in the vermilion border (the vermilion border is the red portion of the lip), a fibrous band or depressed groove running up to the nostril, or a minor deformity of the nose on the same side.

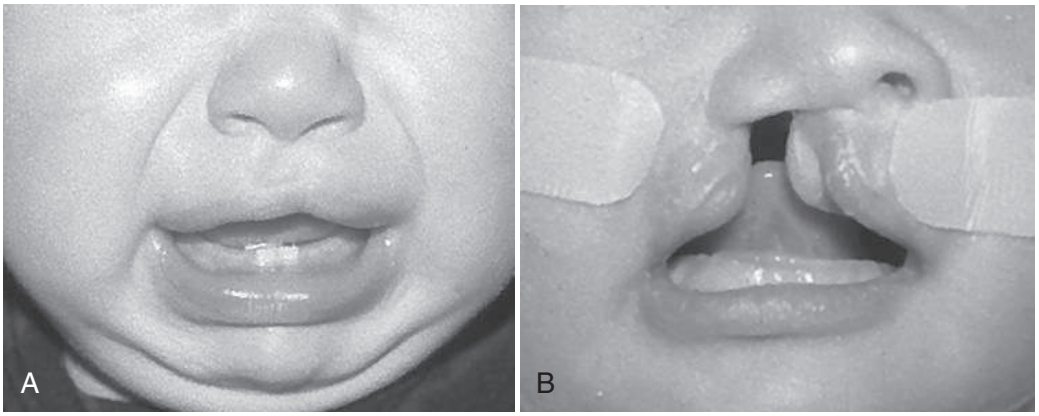

FIGURE 1-1 A unilateral cleft of the lip may vary from a barely detectable microform **(A)** to a complete cleft through the lip and the base of the nose **(B)**. The tape seen on the lateral segments in **B** is there in preparation for a small acrylic device or additional tape that will serve to bring the protrusive premaxilla (*baby's left)* into better position for surgical repair of the lip. (From Peterson-Falzone SJ, Hardin-Jones MA, Karnell MP: *Cleft palate speech* [4th ed]. St. Louis: Elsevier, 2010.)

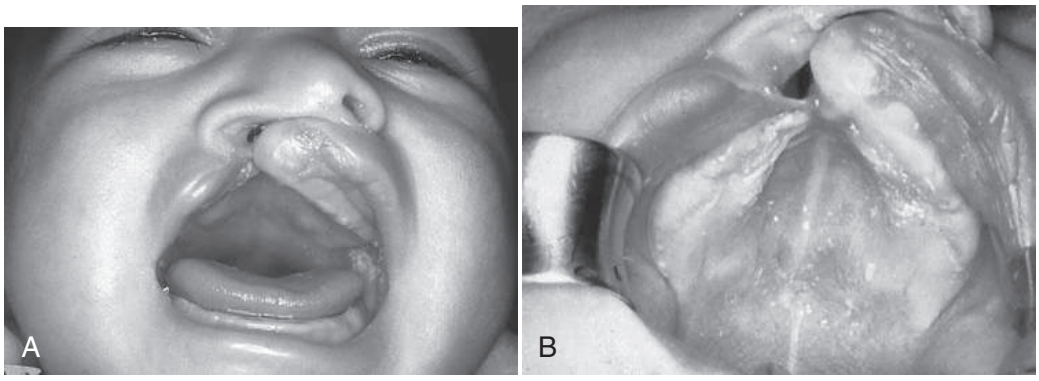

FIGURE 1-2 **A** and **B,** Incomplete cleft of the lip and alveolus on the right, with intact secondary palate. Note flattening of the nose on the cleft side. (From Peterson-Falzone SJ, Hardin-Jones MA, Karnell MP: *Cleft palate speech* [4th ed]. St. Louis: Elsevier, 2010.)

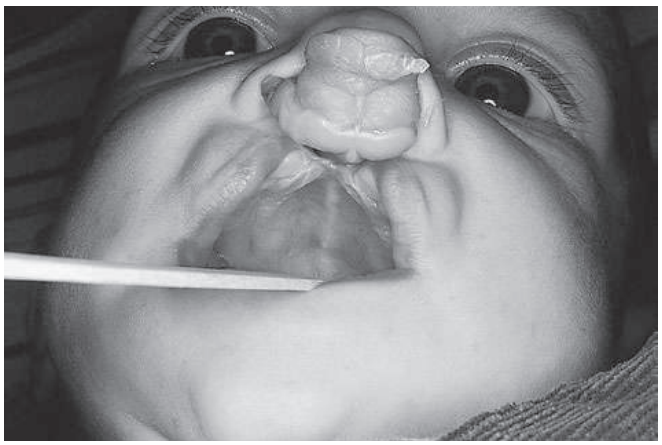

FIGURE 1-3 Infant with complete bilateral cleft of the primary palate but intact secondary palate. (From Peterson-Falzone SJ, Hardin-Jones MA, Karnell MP: *Cleft palate speech* [4th ed]. St. Louis: Elsevier, 2010.)

Minimal lip defects may be associated with a minor deformity of the anterior portion of the maxillary alveolar arch. The term *forme fruste* is often applied to a minor defect of the lip with or without a minimal defect of the alveolus. Minimal defects of the lip and alveolus have no effect on speech, but the patient (and family) may want to seek evaluation by a geneticist/dysmorphologist because the same genes that contribute to complete clefts also contribute to minor defects.

Some further points about cleft lip:

1. Both unilateral and bilateral clefts of the lip can occur without cleft palate (Figs. 1-2 and 1-3).
2. Bilateral clefts of the lip are often asymmetrical, with one side wider or more complete than the other (Fig. 1-4).
3. If there is a complete bilateral cleft of the lip, the central portion of the lip and alveolus (known as the *premaxilla*) is attached directly to the tip of the nose, with little or no columella (Fig. 1-5). In these cases the premaxilla is very protrusive (Fig. 1-6).

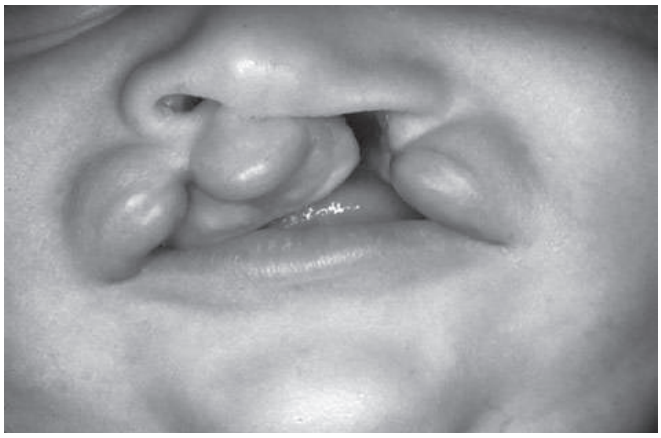

FIGURE 1-4 Asymmetrical bilateral cleft lip, complete on the baby's left side but incomplete on the right. (From Peterson-Falzone SJ, Hardin-Jones MA, Karnell MP: *Cleft palate speech* [4th ed]. St. Louis: Elsevier, 2010.)

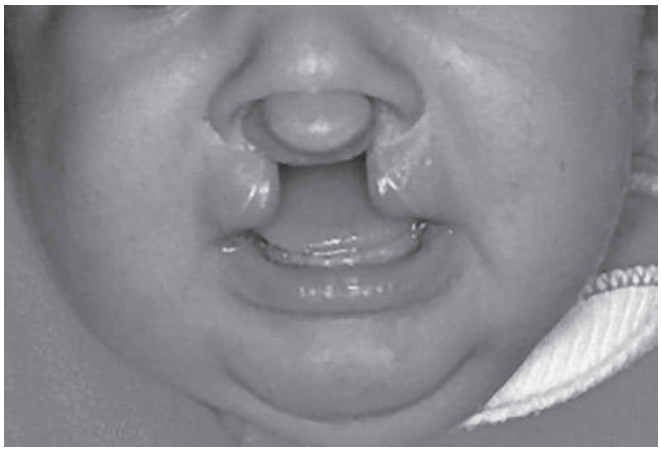

FIGURE 1-5 Bilateral cleft lip and alveolus with the central portion of the upper lip and alveolus attached to the tip of the nose with little or no columella. (From Peterson-Falzone SJ, Hardin-Jones MA, Karnell MP: *Cleft palate speech* [4th ed]. St. Louis: Elsevier, 2010.)

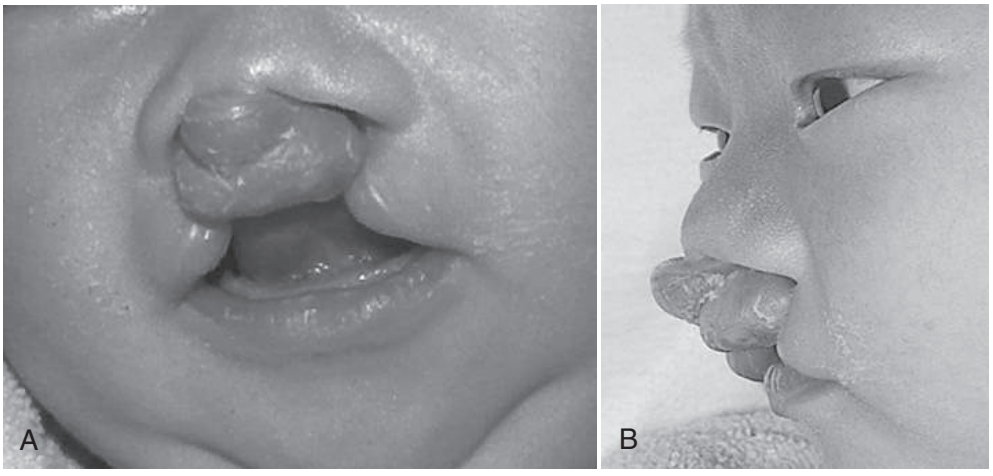

FIGURE 1-6 **A** and **B,** Two views of the protrusive premaxilla in an infant with complete bilateral cleft lip. (From Peterson-Falzone SJ, Hardin-Jones MA, Karnell MP: *Cleft palate speech* [4th ed]. St. Louis: Elsevier, 2010.)

4. Unilateral clefts of the lip or lip and palate are almost twice as likely to occur on the left side than on the right.
5. Even complete clefts of the lip should have no effect on speech, once the clefts are repaired. The exception to this may be a poor repair resulting in a lip that is deficient in length (see Chapter 3 on physical management). However, the dental problems that often occur in association with cleft lip and alveolus (missing and malpositioned teeth) may contribute to articulation problems.

CLEFT PALATE

Clefts of the secondary palate also range from minimal defects to complete open (overt) clefts extending all the way from the uvula (or what would have been the uvula) forward to the region of the incisive foramen. They vary greatly in width (Fig. 1-7).

A defect of the secondary palate may be as small as a slight scallop or streak in the uvula (Fig. 1-8). The findings on the clinician's intraoral examination may vary from these slight differences in the appearance of the uvula to a cleft through the uvula and partly into the velum to a complete cleft of the velum. Overt clefts of the velum that do not reach all the way forward to the incisive foramen still usually have a submucous extension (division of the musculature) anterior to the open portion.

Some further notes about clefts of the secondary palate:

1. Small defects in the velum are often missed in the immediate postnatal period unless the baby exhibits feeding problems.
2. Very large clefts of the secondary palate can leave the surgeon very little tissue to work with (see Fig. 1-7, *J*).
3. In very large clefts of the palate, the base of the vomer bone can usually be seen in the midline, and it is very straight because it is not attached to either

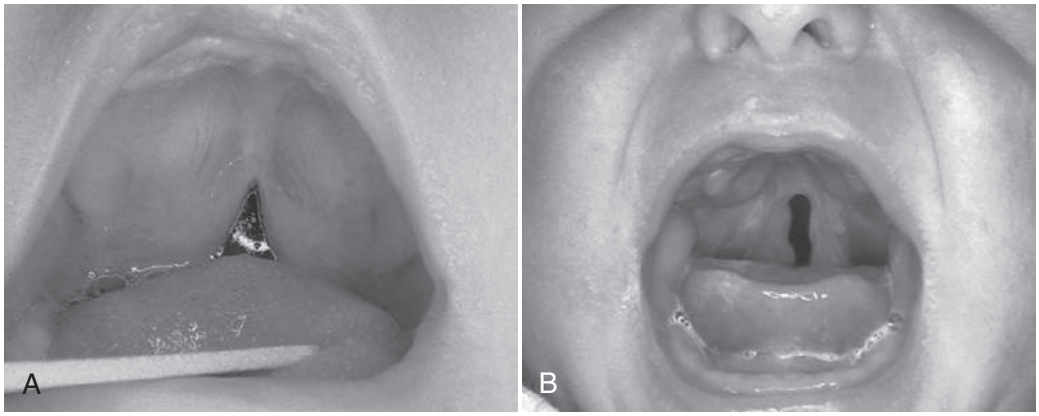

FIGURE 1-7 Wide range in severity of clefts of the secondary palate. **A** to **J,** The range also includes, on the lesser end, the clefts shown in Figure 1-8 and even smaller, less easily detectable (and occult) clefts of the secondary palate. In **B, D, F,** and **H,** note that deformity of the nose is not usually seen in patients with clefts of the secondary palate only. (From Peterson-Falzone SJ, Hardin-Jones MA, Karnell MP: *Cleft palate speech* [4th ed]. St. Louis: Elsevier, 2010.)

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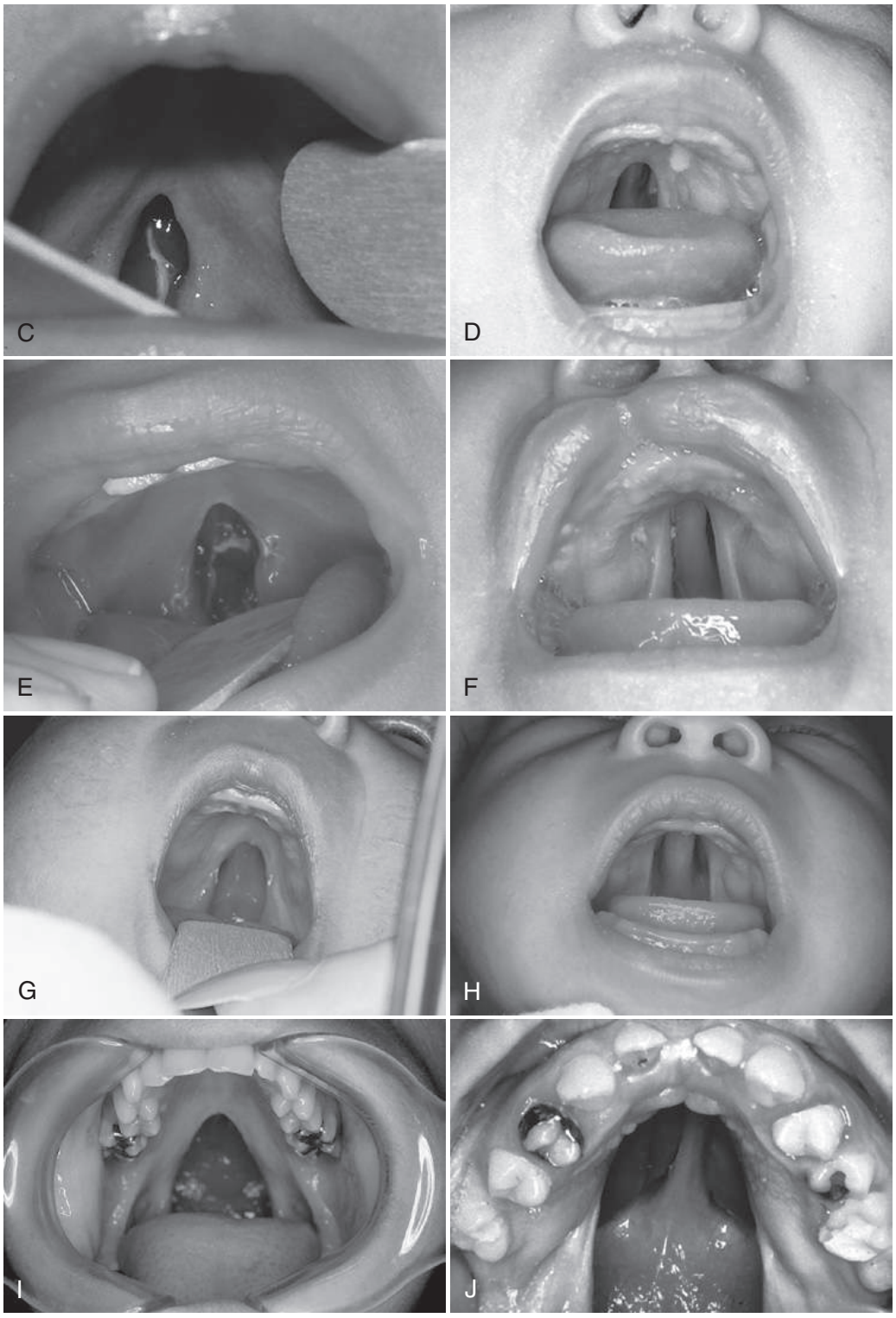

FIGURE 1-7, cont'd

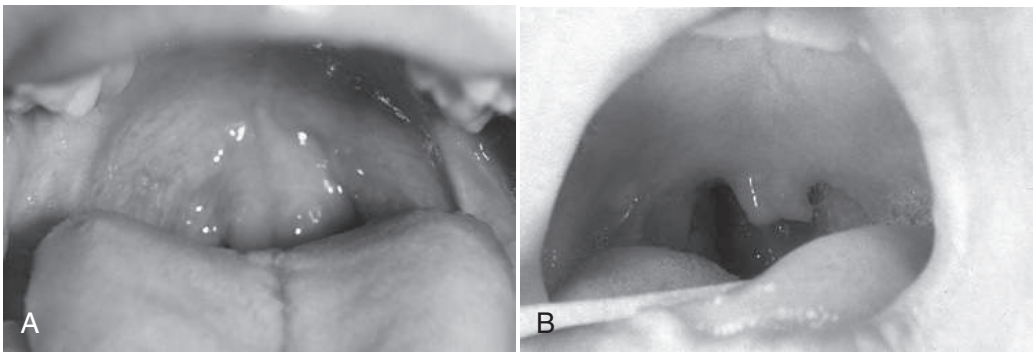

FIGURE 1-8 **A** and **B,** Submucous defects of the secondary palate with a bifid uvula that may escape detection on a quick or careless intraoral examination. These photographs illustrate what may be considered microforms of clefts of the secondary palate, but it is important to remember that the patients may be either asymptomatic in speech or exhibit significant velopharyngeal inadequacy. The intraoral view alone tells us little of clinical significance, but the presence of the bifid uvula is important for genetic documentation of each case regardless of whether that person is symptomatic in speech. (From Peterson-Falzone SJ, Hardin-Jones MA, Karnell MP: *Cleft palate speech* [4th ed]. St. Louis: Elsevier, 2010.)

palatal shelf. (Thus there is no "tug" to one side.) Technically, this may be termed a "bilateral cleft of the secondary palate," but such terminology is confusing to clinicians who look in the mouth and see only one cleft, not two.

4. The patient with cleft lip and palate can have a significant discrepancy between the size of the lip defect and the size of the palatal defect (Fig. 1-9).

Submucous Clefts of the Secondary Palate

Submucous clefts of the secondary palate may occur with no actual opening into the nasal cavity, or they may be combined with a partial posterior opening in the velum. Like actual open clefts, submucous clefts vary in extent or severity. For example, the dehiscence (split) in the musculature of the velum may be narrow or wide.

In a submucous cleft of the velum, the muscle fibers that should have reached across the midline to interlock with those from the other side of the developing palate instead run forward. This is not really a division in the musculature but muscle running in the wrong direction.

Submucous clefts with central perforations through the thin mucosa have also been described in the literature, often with the label of "congenital palatal fistulas." See the note on page 18 of Peterson-Falzone et al. (2010).

The three classic visible stigmata of a submucous cleft are bifid uvula, muscular dehiscence of the velum (sometimes termed a "zona pellucida"), and a bony defect of the hard palate (Fig. 1-10). However, here are some warnings about this "classic" description:

1. The three stigmata do not always appear together; one may be present without the other two, or two may occur without the third. A patient may have only

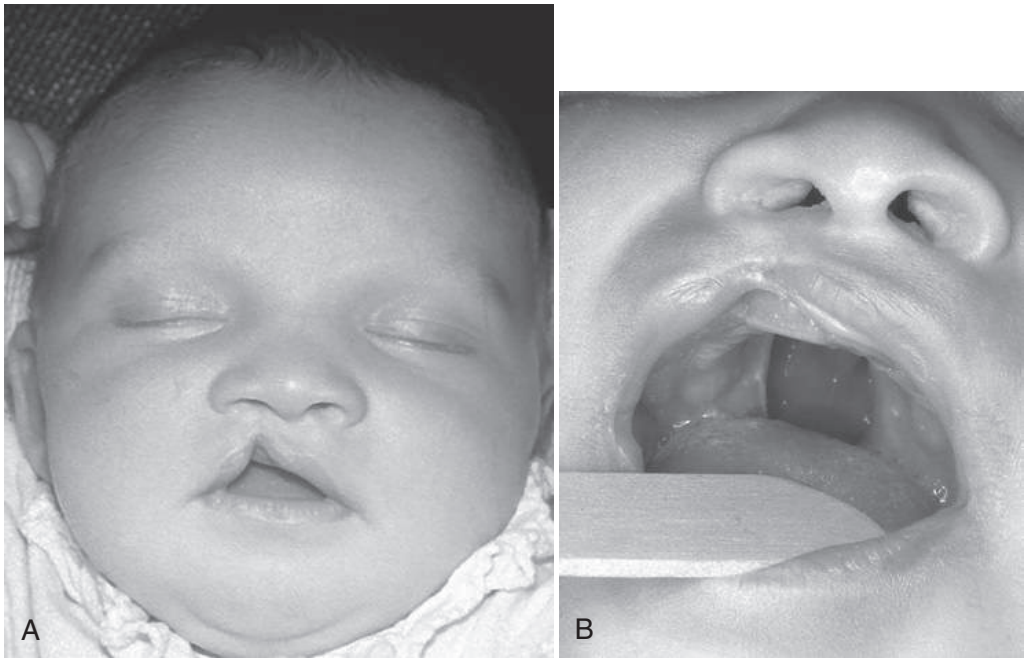

FIGURE 1-9 Facial **(A)** and intraoral **(B)** views of an infant with an incomplete unilateral cleft, in that the lip is only partially cleft **(A),** but there is nevertheless a wide cleft of the palate **(B).** (From Peterson-Falzone SJ, Hardin-Jones MA, Karnell MP: *Cleft palate speech* [4th ed]. St. Louis: Elsevier, 2010.)

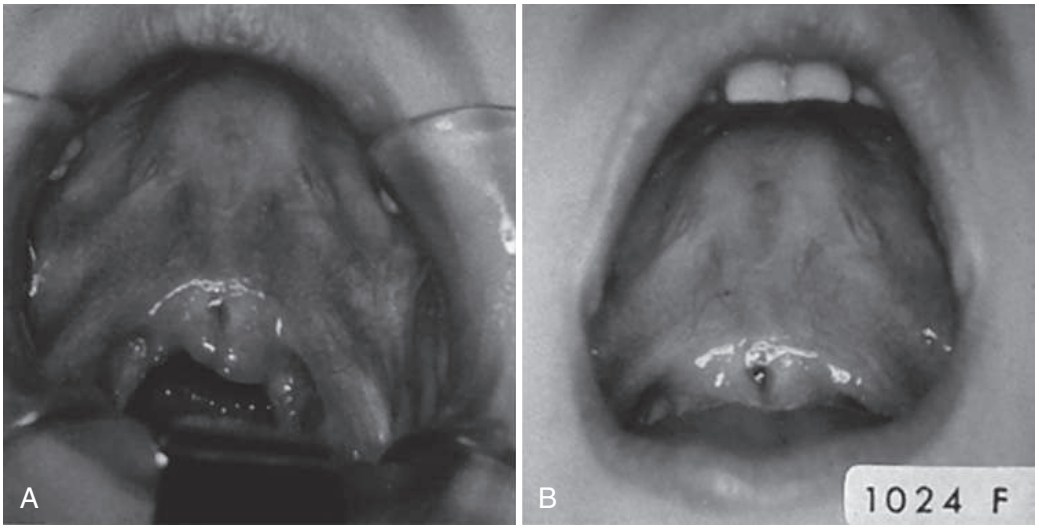

FIGURE 1-10 **A** and **B,** Bifid uvula, zona pellucida (transparent area) of the soft palate (difficult to see in a black and white photograph), and submucous notch in the posterior border of the hard palate. (From Peterson-Falzone SJ, Hardin-Jones MA, Karnell MP: *Cleft palate speech* [4th ed]. St. Louis: Elsevier, 2010.)

a small defect in the uvula or a bifid uvula and muscular diastasis of the soft palate with an intact hard palate.

2. Even when none of the three classic stigmata is visible from the intraoral view, the patient may have a midline trough-like defect on the upper surface of the soft palate that can be seen only on nasendoscopy. This is often labeled an "occult submucous cleft."
3. Although nasendoscopy has been a routine clinical examination since at least the 1980s, only a relatively small percentage of cases of submucous cleft palate reported in the extensive literature on this subject have undergone a nasendoscopic examination. Thus it cannot be known how often one clinical sign of a submucous cleft appears in conjunction with others.
4. In the older literature, patients were sometimes assumed to have submucous clefts simply because they had hypernasal speech. In addition, in some reports patients with other anatomical or neurological findings affecting velopharyngeal (VP) function were grouped with patients with submucous clefts. This practice negated the validity (and value) of many reports.
5. Keep in mind that some speakers have all three classic visible stigmata of a submucous cleft but do *not* have impaired velopharyngeal function. We do not know how many such individuals there are because we do not learn about them if they do not have problems. It is good to remember that no one-to-one correlation exists between the visible signs of a submucous cleft and the presence or absence of audible signs of VP inadequacy (VPI).

"Noncleft" Problems in Velopharyngeal Function

Some individuals have speech characterized by the perceptual characteristics of "cleft palate speech" but without an overt cleft or any of the signs (overt or covert) of a submucous cleft. As discussed in detail in Chapter 5, there are multiple etiologies for noncleft VPI—some structural, some neurological, and some behavioral. It is critical that an experienced speech-language pathologist be at the helm of the decision-making team that identifies the etiology and makes the appropriate plan for treatment. In the past, children were too often subjected to months or years of essentially useless speech therapy without any of the caregivers first trying to look at the movement of the VP system during speech (with a videoendoscopic or videofluoroscopic study) or even obtaining a still, lateral x-ray study of the head to look at the size and relationship of the relevant structures. Chapter 5 provides all the details on causes of noncleft VPI, and Chapter 12 contains the pertinent information on therapy.

WHY CLEFTS OCCUR (ETIOLOGY)

Although speech-language pathologists do not serve as genetic counselors for families, they may be very curious about why clefts occur. Both CL ± P and CPO are, in general, caused by a combination of factors, neither entirely genetic nor entirely environmental in most cases. Both conditions are etiologically heterogeneous. In a 2004 tutorial on this subject, Marazita and Mooney (2004) concluded (from their own work and that of others) that various genetic factors cause clefts in 20% to 50% of the cases, and the remaining clefts are attributable to either