



CHAPTER 2

Speech: Something We Can Really Fix

As we have just seen, speech impairment is one of the most common findings in VCFS, occurring in at least 70% of cases (Shprintzen & Golding-Kushner, 2009).

In this chapter, we discuss the sequelae, diagnosis, and management of velopharyngeal insufficiency (VPI), the disorder that leads to hypernasality. Articulation and language impairment are discussed in terms of their interrelationship with VPI because each of these components is affected in some way by inability to close the velopharyngeal valve during speech directly or indirectly.

IMPAIRMENTS OF VERBAL COMMUNICATION IN VCFS

As mentioned in Chapter 1, when most people think of “speech,” they typically think of the entirety of verbal communication, but the act of speaking to someone else in order to communicate has multiple components. Individuals with VCFS are prone to impairments of all the components of verbal communication. These components include:

Articulation: the production of individual sounds using the tongue, lips, teeth, palate, larynx, and pharynx.

Voice: the production of sound at the level of the vocal folds.

Resonance: the modification of the sound created by the vocal folds by having it resonate in the chambers above the vocal folds (the pharynx, oral cavity, and nasal cavity).

Together, articulation, voice, and resonance combine to create the speech signal and are the three functions that comprise speech. These are the motor aspects of oral communication, and together with fluency, are the aspects of speech we hear produce and hear.

Language: the cognitive aspect of communication; it is the communication of thoughts, information, ideas, or feelings through a system of signs, sounds, gestures, or written marks that are fundamentally arbitrary but are rule-governed and socially shared.

SPEECH SYMPTOMS OTHER THAN HYPERNASALITY: RELATIONSHIP TO VELOPHARYNGEAL INSUFFICIENCY

Articulation



Although hypernasality is severe in VCFS and at the root of the compensatory articulation patterns that occur in most children with VCFS, it is the articulation disorder that most severely impairs speech intelligibility. If articulation is correct, hypernasality, even when severe, typically does not prevent speech from being understood. The glottal stop substitution pattern that occurs in a high percentage of children with VCFS severely limits intelligibility because it reduces the number and range of consonants produced during speech production. Even if surgery successfully eliminates velopharyngeal insufficiency and hypernasality, if the articulation impairment remains, speech will sound severely impaired and unintelligible (Video 2-1). Therefore, it is essential to correct the articulation impairment separately. Furthermore, it is necessary to correct it separately, because surgical correction of VPI has no effect on compensatory speech errors. Surgery can only correct hypernasality and one class of speech errors that are obligatory, nothing more. In fact, defining success of surgery based on speech intelligibility, or “speech improvement,” has no relevance to what surgery is designed to do.

The earlier these abnormal articulation patterns are detected, the easier they are to correct. It is even better to prevent them before they develop. Prevention and treatment of the articulation impairment in VCFS are described in detail in Chapter 3

In all languages on the planet, the majority of our speech sounds are produced without nasal resonance. Consonant sounds, the sounds that carry nearly all of the meaning in our speech, are almost all produced by pressure created in the oral cavity, and vowel sounds resonate in the oral, not the nasal, cavity. In English, only three consonants have nasal resonance; the /m/ sound, the /n/ sound, and the /ŋ/ (phonetically transcribed as /ŋ/). All of our other consonants, such as /b/, /p/, /t/, /d/, /s/, /z/, /f/, /v/, /k/, /g/, the /θ/ (the th in “math”) and /ð/ (the voiced th in “the”), the /dʒ/ (the j in “jump”), and the /g/ sound in “go,” /ʃ/ (sh), and /tʃ/ (ch), and the sounds /r/, /l/, /h/, /j/, (the y in

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“you”) and /w/, are produced without any sound or air coming out of the nose in normal speech production. When VPI is present, there is reduced air pressure in the mouth and sound escapes through the nose. If the reduction in oral air pressure is severe, attempts to produce sounds will result in production of the nasal cognate of the sound. Thus, /b/ would sound like /m/. This is because the sounds are both produced by closing the lips, but the key difference is that oral air pressure is built up and then released for /b/ (thus requiring VP closure) but /m/ is produced with no oral air pressure, just with air escape through the nose (thus not requiring VP closure). At an extreme of VPI, the consonant sounds that normally have no nasal resonance are produced abnormally in one of several ways. At best, with good articulation and some oral air pressure, they would be produced correctly but with some nasal air escape through the nose that might be audible. If VPI is severe, the consonants produced with good articulation but minimal air pressure would sound like /m/, /n/, or /ŋ/, the three nasal phonemes. At worst, the child would abandon attempts to produce the articulation sounds in the mouth and substitute a glottal stop or other compensation, described in Chapter 1.

It should be pointed out that hypernasal resonance is heard on vowels. It is a vowel event. Nasal air escape (nasal emission) occurs on consonants, as does nasal turbulence. One of the unique characteristics of the /m/, /n/, and /ŋ/ sounds is that, unlike other consonants, they are produced with nasal coupling and, therefore, have nasal resonance. They are continuant sounds (can be prolonged) and voiced (the vocal folds continue to vibrate to make voice while the lips and tongue modify the sound produces). Other consonant sounds, referred to as “stops” or “plosives,” stop the air (like /p/, /b/, /t/, /d/, /k/, and /g/). Still others, the “fricatives,” constrict the outgoing airstream, thus creating friction sounds, add hissing noises to the oral airstream (like /s/, /f/, and /θ/).

Timing of velopharyngeal closure also is significant. In normal velopharyngeal closure, at the beginning of an utterance, the palate elevates and the lateral pharyngeal walls begin to close the velopharyngeal valve before the first sound is produced unless the first sound in the utterance is a nasal consonant or a nasalized vowel. Once velopharyngeal closure is achieved, the valve stays shut until the utterance is completed or a nasal consonant is produced (Video 2-2). Note that closure occurs even on the vowels as long as they are surrounded by two nonnasal consonants. In some people who sound very hypernasal, but seem to produce normal consonant pressure for plosives, fricatives, and sibilants, closure is achieved on most or all nonnasal consonants, but they open the valve every time a vowel is produced. The type of “pulsing” of the velopharyngeal valve is perceived as consistent hypernasality because the vowels are where we judge resonance (Video 2-3). Therefore, it is critically important for a child who is perceived to be hypernasal to have studies performed to directly view the velopharyngeal mechanism and determine if VPI is consistent, or if there are timing issues in VP closure causing

resonance to be perceived as hypernasal. The assessment of hypernasal speech is dependent on understanding the specific phenotypic features of VCFS that can affect the functioning of the velopharyngeal valve and implementing procedures in an age-appropriate manner so that the maximum information is obtained.

Factors Contributing to Velopharyngeal Insufficiency in VCFS

People with VCFS typically show severe hypernasality caused by a number of factors:

1. ***Overt, submucous, or occult submucous cleft palate.*** Palatal anomalies are common in VCFS, occurring in approximately 70% of affected individuals (Shprintzen & Golding-Kushner, 2008). Shprintzen et al. (1985) found that, among 1000 individuals with cleft palate and/or hypernasal speech, 8.1% had VCFS. The largest percentage of these cases had occult submucous cleft palate or submucous cleft palate. Overt clefting is the least common form of palate anomaly in people with VCFS (Shprintzen & Golding-Kushner, 2008). All of these palatal anomalies reduce the muscular composition of the velum so that movement, thickness, and length are all deficient (Golding-Kushner, 1991; Zim, Schelper, Kellman, Tatum, Ploutz-Snyder, & Shprintzen, 2003), increasing the likelihood of velopharyngeal insufficiency.
2. ***The pharyngeal and palatal muscles are hypotonic and move poorly.*** In their initial description of VCFS, Shprintzen, Goldberg, Young, and Wolford (1978) reported absent movement in the velum and lateral pharyngeal walls for the 12 patients described. In a follow-up report (Shprintzen et al., 1981), pharyngeal hypotonia was reported to be a consistent finding in VCFS and this has been confirmed since that time (Golding-Kushner, 1991; Shprintzen & Golding-Kushner, 2008). Therefore, VPI tends to be severe with little or no closure of the valve during normally nonnasal speech. This allows most of the resonating sound to escape into the nasopharynx and nasal cavity. In fact, Zim et al. (2003) found that the muscle tissue of the velopharyngeal region was histologically abnormal in children with VCFS, with abnormalities noted in the type and quantity of muscle fibers.
3. ***The nasopharynx is larger in VCFS than in other people of the same size and age.*** This increase in size of the nasopharynx has multiple components including smaller than normal adenoids (Williams, Shprintzen & Rakoff, 1987) (Figure 2-1), a flat skull base (platybasia) that increases the depth and volume of the nasopharyngeal airway (Arvystas & Shprintzen, 1984; Golding-Kushner, 1991) (Figure 2-2), and decreased

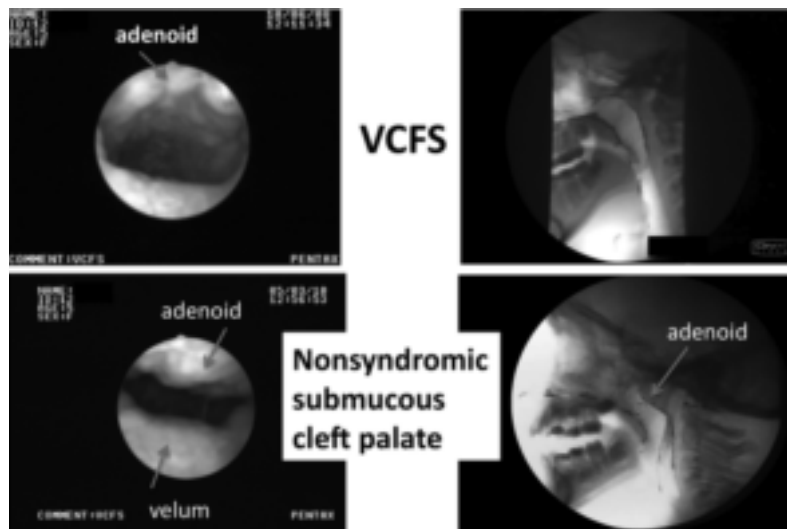


FIGURE 2–1. Nasopharyngoscopy and lateral view videofluoroscopy in a 5-year-old female with VCFS (*top row*) compared to a 5-year-old female with nonsyndromic submucous cleft palate (*bottom row*). Note the near absence of adenoid in the VCFS case that increases nasopharyngeal volume compared to the case with nonsyndromic submucous cleft.

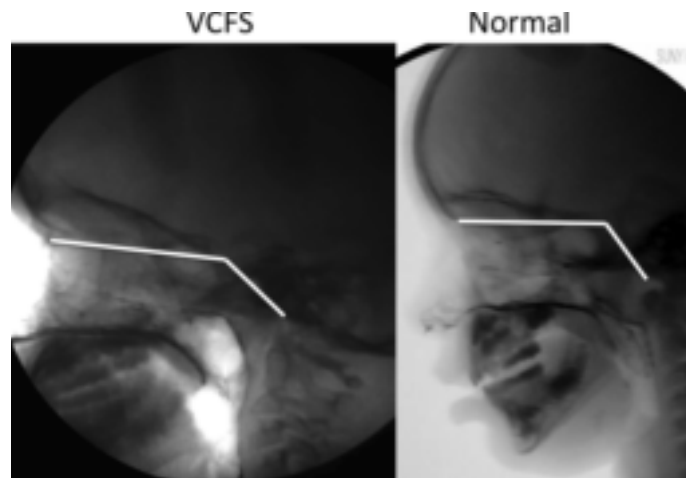


FIGURE 2–2. Obtuse cranial base angle in a child with VCFS (*left*) compared to a normal child (*right*) as seen in a lateral view radiograph.

thickness of the pharyngeal walls (Golding-Kushner, 1991) (Figure 2-3). The decreased thickness of the muscle wall is consistent with reduced muscle mass and therefore the likely reduction in pharyngeal movement. Adenoid size has been found to be strongly related to the presence or

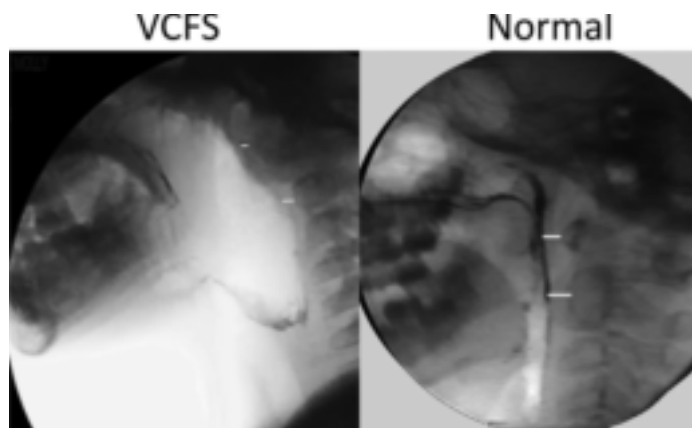


FIGURE 2–3. Decrease in posterior pharyngeal wall muscle thickness in VCFS (*white lines*) compared to a normal child of the same age and sex.

absence of velopharyngeal insufficiency in VCFS. Children with VCFS and small adenoids rarely have normal speech whereas some of those with larger adenoids develop speech without hypernasality (Golding-Kushner, 1991; Havkin, Tatum, & Shprintzen, 2000).

4. ***Structural and functional asymmetry of the pharynx and palate.*** A high frequency of asymmetry of the velum and the pharynx in individuals with VCFS has been reported (Chegar, Tatum, Marrinan, & Shprintzen, 2006). Specifically, 67% of children with VCFS who had velopharyngeal insufficiency had asymmetric elevation of the palate during speech when velar motion was present. Structural pharyngeal wall asymmetry was found in 84%. When compared to an age-matched normal control sample, significant differences were found in structure and function of the pharynx and palate in individuals with VCFS. Asymmetry of movement and structure of the palate and pharyngeal walls was found to be a frequent contributor to velopharyngeal insufficiency in individuals with VCFS.
5. ***Behavioral affect.*** Children with VCFS often are withdrawn and unsure of themselves. They frequently turn their faces downward, bury their chins in their chests, and use a very low volume voice. This enhances nasal resonance by preventing the child from projecting his or her voice sufficiently through the oral cavity (Video 2–4). Resonance can be affected by effort and voice projection.



DIAGNOSTIC PROTOCOL FOR VELOPHARYNGEAL INSUFFICIENCY

The diagnostic protocol for velopharyngeal insufficiency includes a comprehensive speech-language evaluation, flexible fiberoptic nasopharyngoscopy,

multiview videofluoroscopy, and in cases where pharyngeal flap is being planned, magnetic resonance angiography (MRA) because of anomalies of the internal carotid arteries. In cases where magnetic resonance studies cannot be done, for example, if the patient has magnetic metal somewhere in the body (such as rods, plates, screws, or wires from previous operations), CT angiography may be substituted. We also recommend appropriate medical tests as part of the overall comprehensive evaluation as detailed in Chapter 4 of Volume I, but these are part of a patient's complete evaluation before he or she is considered for surgery. We never proceed with surgery for VPI in individuals with VCFS without these studies. The protocol is the same as we recommend for patients who do not have VCFS but who have cleft palate or resonance disorders. The exception is MRA, which is done only for patients with VCFS. To date, we have not encountered other syndromes or people in the general population who have similar anomalies of the internal carotid arteries.

Speech-Language Evaluation

This evaluation needs to be conducted by a clinician who is familiar with disorders associated with resonance imbalance, compensatory articulation, VPI, and preferably VCFS as well. Not all speech pathologists are familiar with these disorders and may not understand the basic anatomy, physiology, and developmental issues and that can lead to serious errors in judgment about the appropriate course of treatment. The diagnostic protocol should include a comprehensive standardized articulation test, measures of language development; analysis of spontaneous language in conversation (if age appropriate); a clinical assessment of resonance (by listener judgment); a clinical assessment of nasal air escape during speech that might include the use of a mirror, stethoscope, or listening tube; a clinical assessment of voice (i.e., hoarseness and issues related to laryngeal function, pitch, and volume); and a few minutes of diagnostic therapy to demonstrate and achieve the correct production of sounds in the place of the compensatory patterns. This is sometimes referred to as "stimulability testing," in other words, trying out some procedures to determine the most effective in a short time. The specific articulation and language tests to be used can be debated among clinicians and researchers. Many are available. Language evaluation instruments that have been used to assess children with VCFS are discussed by Scherer et al. (1999) and can be added to standard batteries of language instruments already in use by clinicians. A useful language battery for VCFS might include the CELF language battery (*Clinical Evaluation of Language Fundamentals*), the TTC (*The Token Test for Children*), and the *Structured Photographic Expressive Language Test* (SPELT) (see Appendixes). The articulation tests can include broad instruments such as *The Goldman-Fristoe Test of Articulation* or tests specific to children with VPI such as *The Great Ormond Street Hospital Cleft Audit Protocol for Speech* or the *Iowa Pressure Articulation Test*. The test should elicit production of all phonemes in all positions of words. It is impor-

tant to note if there is any variation in resonance or nasal air emission with different sounds or sound groups. On occasion, complete velopharyngeal closure can be achieved for some sounds and not others. In such cases, this information would be important to know before endoscopic and fluoroscopic assessment so that those sound variations can be explored in detail during the instrumental portions of the examination. Additionally, those variations may indicate that the most appropriate treatment is speech therapy rather than surgery. These issues are explored in detail in Chapter 3.

Instrumental Examinations

Although a wide variety of instruments and techniques are available for assessing velopharyngeal function, our approach is to use only techniques that provide direct visualization of all of the elements of velopharyngeal function during the unimpeded flow of connected speech. Only two techniques today do this in real time and these are flexible fiberoptic nasopharyngoscopy (also referred to as nasal endoscopy) and multiview videofluoroscopy. We do not use the indirect measures of velopharyngeal function, such as nasometry or pressure flow studies for diagnostic purposes. Our preference is to rely on our ears to judge resonance and nasal air emission because they are an issue only if they are heard. Both nasometry and pressure flow have been described as being “objective” instruments because they provide a numerical measure from their circuitry. However, a quote attributed to the late Albert Einstein says, “Not everything that counts can be counted and not everything that can be counted counts.” The only relevance of a resonance disorder is what we hear and can describe as being abnormal, not a number on a machine unless it can be demonstrated that this number has relevance (i.e., validity) in the diagnostic and treatment process. The purpose of listener judgment (perceptual assessment) is to decide that treatment for hypernasality is needed. Once that decision has been made, then the information that is required related to how to physically manage the velopharyngeal insufficiency. Although indirect measures such as nasometry or pressure-flow can provide a statistic to correspond to the “size” of a velopharyngeal gap, it cannot tell the surgeon anything about the shape or location (central or skewed to one side, or vertical position) of the gap. Thus, it gives the surgeon no information to assist in planning surgery. It also does not give the SLP information on variations related to articulatory precision or variation. Thus, the indirect measures duplicate the information you can get in a very low-tech manner (listening) and do not provide guidance for treatment. The planning of intervention and treatment of VPI requires direct visualization of the size, shape, location, and timing of opening and closing of the VP region through which communication of air and sound into the nasopharynx occurs. In 1990, the International Working Group (Golding-Kushner et al., 1990) recommended that the optimal instrumental assessment of velopharyngeal function should utilize the combination of nasopharyngoscopy and videofluoroscopy in at least

two views, lateral (midsagittal) and frontal (P-A or posterior-anterior). Our preference in performing these procedures for assessing velopharyngeal function is to perform them after the age of 4 years when it is possible to obtain adequate compliance with endoscopy and fluoroscopy to provide adequate diagnostic information for the recommendation of surgical procedures. In addition to better compliance with the procedures, language production usually is adequate to obtain reasonable speech samples for assessment by that age.

Nasopharyngoscopy

In most centers, endoscopic assessment of the velopharyngeal valve has become the gold standard for visualization procedures. Although highly valuable, endoscopy is highly dependent on the skill of the endoscopist in terms of the technical aspect of obtaining the study and in terms of interpreting the images. Endoscopy provides a direct view of the velopharyngeal valve from above during unimpeded speech because the instrument is passed through the patient's nose, not the mouth. Key to the procedure in young children, particularly children with VCFS, is the ability to make the examination painless so that complete cooperation is enlisted from the patient. Although this may sound obvious, it is extremely important for the examiner to be good with children in order to gain their compliance. Things should be explained to them in terms they will accept and understand. Children should be assured that they will not be harmed during the examination. The nose should be well anesthetized, and the endoscope should be entered into the nose using visual guidance so that the examination is painless. Although there will always be some children who are difficult to examine, it is possible to gain cooperation from the large majority of children over 4 years of age. Most children will be very nervous as the examination begins because they are not accustomed to having anything put into their nose (at least not deeper than they place their own fingers), even if the device is thinner than a finger.

One extremely important point: **all examinations must be recorded in some type of video format (preferably digital video) for subsequent review.** Examinations that are not recorded are, for all practical purposes, useless. Careful phonetic analysis often demands viewing the examination multiple times. In addition, the recording is needed so the surgeon can review the pattern of closure before surgery without having to repeat the endoscopy, which may have been done weeks prior. The examination may need to be reviewed by multiple people working with the child, including the SLP, ENT, and surgeon. They may not all be present at the evaluation, or may want to show the examination to a colleague. A video of the examination also serves as an important element of the medical record for comparison with additional studies that may be done in the future.

The procedure is conducted as follows (Video 2-5). The child is seated in facing the examiner who is also seated, so as not to loom over the child,



which can be a bit scary. The examiner assures the child that the procedure will not be painful and will not harm the child. The examiner then explains everything that will happen before and during the procedure to the child, including what it might feel like, if the child feels anything at all. The examiner should make certain that the child is paying attention by redirecting the child when necessary. The preferred setup for the endoscopy is to have a video display facing the examiner and a second display behind the examiner facing the child (Figure 2-4). In this way, the child can see the endoscopy and it will help to fix their gaze and attention. Following the explanation, the examiner anesthetizes the nose topically. The larger nostril is selected initially by direct vision followed by listening to the airflow through each nostril by occluding the opposite nostril and having the child sniff in or blow out. The lower the pitch, the more patent the airway. Once the larger side is identified, topical anesthesia is applied to that side. Our preferred procedure for achieving topical anesthesia that has been used on more than 10,000 occasions over the past 30 years is to take a piece of cotton packing cut from a strip of Webril undercast padding (Figure 2-5) cut into a thin strip approximately 3-mm wide. That strip is then twirled into a narrower strip and doused with 2% Pontocaine (aqueous tetracaine hydrochloride). Pontocaine is preferred because it is odorless and tasteless and achieves adequate topical anesthesia within minutes. Using a pair of small bayonet forceps, the cotton packing is slid deeply into the nose and left in place for approximately 3 minutes (Video 2-5). It is important to be sure the packing goes deep in the nose and does not bunch in the front, so as to apply the anesthesia to the posterior edge of the nostril. The packing then is removed with the forceps and the endoscope is inserted.

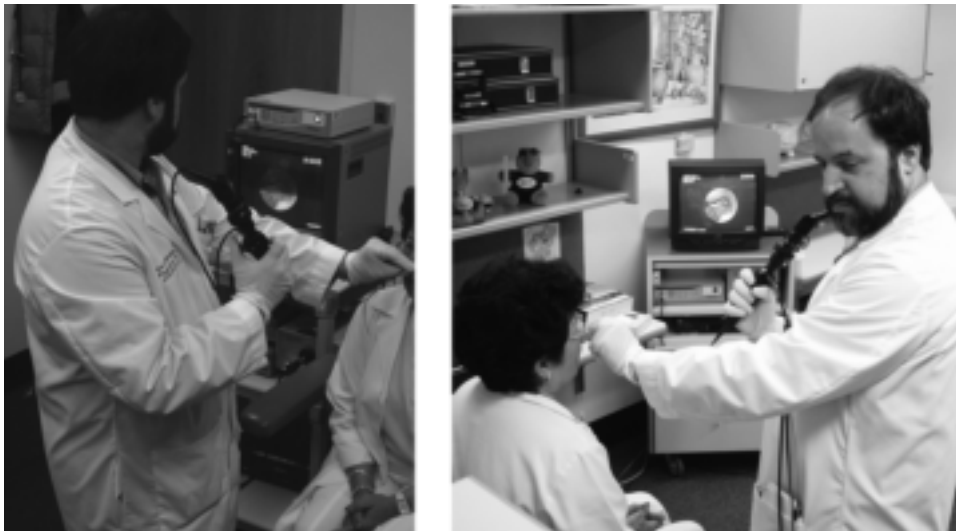


FIGURE 2-4. Arrangement of nasopharyngoscopy equipment allowing the patient to observe the study at the same time the examiner is viewing the study.

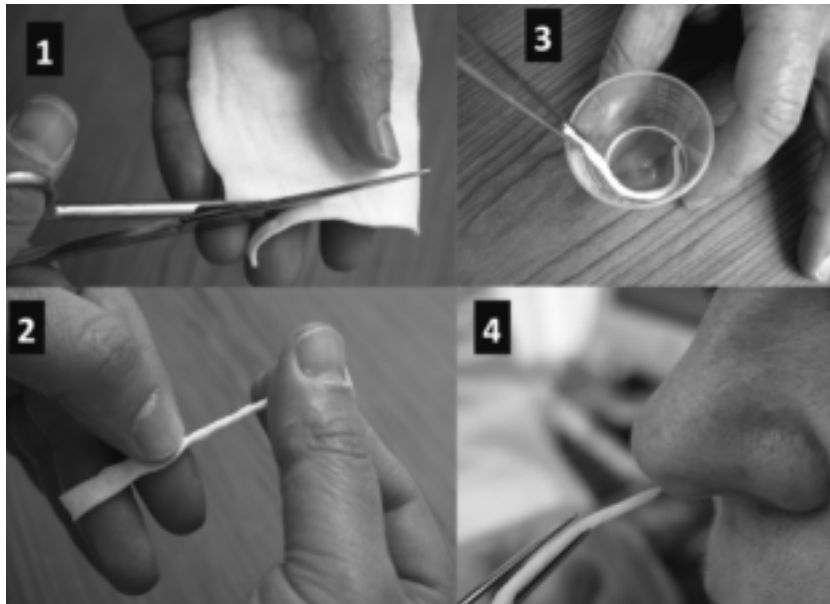


FIGURE 2-5. Preparation of topical anesthesia for examination. A thin strip of Webriil® cotton orthopedic bandage is cut (1) and twirled into a thin packing (2) that is dampened with Pontocaine (2% aqueous tetracaine hydrochloride) and inserted into the large side of the nose with bayonet forceps.

The cotton packing approach is preferred to spray for two reasons. First, the degree of topical numbing of the mucosa is superior using this technique compared to a spray. Second, spraying anesthetic into the nose may allow the anesthetic to be inhaled deeper into the pharynx and even the larynx. Although the effect of potentially anesthetizing the mucosa of the entire upper airway is not known, it is possible that it may alter the outcome of the examination. We also find that children with VCFS often are frightened by the sound and feel of a forceful spray, especially if the atomizer of the type typically found on otolaryngology examination units that have built-in compressors.

Once topical anesthesia is obtained, the endoscope should be passed through the middle meatus of the nose to obtain the best angle of view above the palate and pharynx (Video 2-6). If the endoscope is passed through the inferior meatus, the endoscope will be resting on the palate and when the palate moves, the endoscope will move (Figure 2-6). In the middle meatus, the endoscope will be well above the plane of movement and more of the velopharyngeal valve will be in the field of view. The size and position of the adenoids should be noted. Most important is the view of the palatal structure to determine if there is a midline muscle mass representing the musculus uvulae (Video 2-7). The patient then should repeat a standardized speech sample that contains passages with no nasal consonants, passages with nasal conso-