Speech Outcomes following Pharyngeal Flap in Patients with Velocardiofacial Syndrome

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Background: Velocardiofacial syndrome is the most common defined disorder associated with palatal insufficiency. The authors' purpose is to evaluate one surgeon's experience with correction of velopharyngeal insufficiency in velocardiofacial syndrome using a tailored pharyngeal flap.
Methods: The authors reviewed the records of all children with velocardiofacial syndrome and velopharyngeal insufficiency who were managed with a pharyngeal flap between 1983 and 2009. Data collected included age at operation, preoperative videofluoroscopic findings, speech outcomes, complications, and need for a secondary operation.
Results: The authors identified 33 patients with velocardiofacial syndrome and velopharyngeal insufficiency who had postoperative speech evaluations. Velopharyngeal insufficiency was diagnosed at a median age of 5 years. Palatal findings were: Veau type I ($n = 4$), overt submucous ($n = 6$), or occult submucous ($n = 23$). Median preoperative lateral pharyngeal wall movement was 22 percent (range, 0 to 90 percent). Successful correction of velopharyngeal insufficiency was achieved in 29 of 33 patients (88 percent). One patient had a medially displaced right internal carotid artery, and evidenced intraoperative bleeding and required a blood transfusion. One patient developed obstructive sleep apnea.
Conclusion: A tailored pharyngeal flap is highly effective for correction of velopharyngeal insufficiency in velocardiofacial syndrome with few complications. (*Plast. Reconstr. Surg.* 127: 2045, 2011.)

First described by Shprintzen et al. in 1978,[1] the estimated incidence of velocardiofacial syndrome ranges from one in 2000 to one in 7000 births.[2,3] It can be sporadic or inherited as an autosomal dominant disorder, and is caused by deletion of band 11.2 on chromosome 22.[4–6] This deletion can be diagnosed by fluorescent in situ hybridization[5,7] and with increased sensitivity by multiplex ligation–dependent probe amplification technology.[8,9] Clinical features include characteristic facies, cognitive or behavioral disorders, hearing loss, conotruncal cardiac malformations, urogenital anomalies, hypocalcemia, immunologic disorders, and musculoskeletal abnormalities.[10–13]

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The reported prevalence of velopharyngeal insufficiency in velocardiofacial syndrome ranges between 32 and 75 percent.[13,14] Approximately 75 percent of patients with velocardiofacial syndrome have either overt cleft palate (20 percent), submucous cleft palate (44 percent), or occult submucous cleft palate (38 percent).[15] Adenoidal hypoplasia and cranial platybasia cause a relatively deep pharynx, which further contributes to velopharyngeal insufficiency.[11,12,16] The most impor-

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tant cause of velopharyngeal insufficiency is generalized pharyngeal hypotonia, resulting in limited palatal and lateral pharyngeal wall movement, and is observed in up to 90 percent of patients.[17] Postverbal resolution of velopharyngeal insufficiency contributes to the high prevalence of compensatory articulation (e.g., glottal stops, laryngeal fricatives, and pharyngeal fricatives), which also diminishes lateral pharyngeal wall movement. This combination of anatomical and physiologic abnormalities makes it difficult to treat velopharyngeal insufficiency in velocardiofacial syndrome. A pharyngeal flap is considered the most effective procedure, with success rates of 78 to 98 percent.[18–21] Other surgical options, including palatoplasty (intravelar muscular retropositioning[22] or double-opposing Z-palatoplasty[23]) and sphincter pharyngoplasty,[16] are reported to be less successful.[24–26] The purpose of this study was to evaluate one surgeon's experience with correction of velopharyngeal insufficiency in velocardiofacial syndrome using a lined pharyngeal flap, constructed based on lateral pharyngeal wall motion and the transverse dimension of the oropharynx.

PATIENTS AND METHODS

After approval by the Institutional Review Board of the Committee on Clinical Investigation, we identified and reviewed the charts of all patients who had a diagnosis of velocardiofacial syndrome and had an operation for velopharyngeal insufficiency. A geneticist evaluated all patients, and the clinical diagnosis was made in conjunction with a positive fluorescent in situ hybridization test for 22q11.2 deletion. The senior author (J.B.M.) performed all operations between 1983 and 2009. Patient charts and operative reports were culled, and data collected included date of birth, sex, age at primary operation, preoperative and postoperative speech assessment, videofluoroscopic results, need for a secondary operation, and perioperative and postoperative complications.

Patients with velopharyngeal insufficiency were evaluated at age 4 years or older by multiview videofluoroscopy before a pharyngeal flap. Lateral pharyngeal wall motion (i.e., percentage of closure by medial movement), velopharyngeal gap size (i.e., small, moderate, or gross), and shape of the velopharyngeal defect (i.e., coronal, sagittal, or circular) were recorded. Enlarged tonsils (2+ or greater) were removed at least 8 weeks before the pharyngeal flap. All patients had preoperative magnetic resonance imaging and magnetic resonance angiography to assess possible medial displacement of the internal carotid arteries.

Operative Technique

The pharyngeal flap is constructed with the child in the Rose position.[23] Slight cervical extension helps straighten and laterally displace the sometimes medially deviated internal carotid arteries.[27] The velum is divided in the midline, approximately halfway to the junction with the hard palate, and trapezoidal nasal mucosal flaps are incised and elevated. A superiorly based flap is elevated off the buccopharyngeal fascia to a level above the soft palate. Flap width (either narrow, medium, wide, very wide, subobstructing) is "tailored" based on lateral pharyngeal wall motion (as documented by videofluoroscopy) and transverse oropharyngeal dimension. The pharyngeal donor site is closed by advancing the lateral mucosal edges, which are sutured to the fascia superiorly and apposed inferiorly. The distal end of the upturned pharyngeal flap is placed on the raw nasal surface of the soft palate and secured with three resorbable horizontal mattress sutures. Then, 12- or 14-French red rubber catheters are placed through the nose and along each side of the flap; however, "lateral port control"[28] is not used. The nasal mucosal flaps are sutured to the base of the flap, apposed to line the raw surface of the flap, and the velum is repaired.

Treatment Outcomes

Patients were followed annually in the cleft lip–cleft palate clinic; the senior author examined all patients. A speech pathologist performed preoperative and postoperative perceptual assessments using the Pittsburgh Weighted Values for Speech Symptoms Associated with Velopharyngeal Incompetence instrument.[29–31] Speech performance was based on three structurally correctable variables: *resonance* (normal, mildly hyponasal, mixed hyponasal/hypernasal, inconsistent mildly hypernasal, consistent mildly hypernasal, or moderately or severely hypernasal), *nasal emission* (absent by mirror examination, visible, audible, or turbulent), and *intraoral pressure* (normal or decreased). All children and families were asked whether speech posed a personal or social problem when talking to others.[32] The speech pathologist provided an overall assessment of velopharyngeal function: (1) *normal* (normal or hyponasal resonance, absence of visible nasal emission by mirror examination, normal intraoral pressure, or no personal/social problems); (2) *borderline sufficient* (inconsistent

mildly hypernasal resonance, visible nasal emission, normal intraoral pressure, or no personal/social problems); (3) *borderline insufficient* (consistent mildly hypernasal resonance, audible or turbulent nasal emission, inconsistent decreased intraoral pressure, or a personal/social problem); and (4) *insufficient* (moderately or severely hypernasal resonance, audible or turbulent nasal emission, decreased intraoral pressure, or a personal/social problem). Normal or borderline sufficient velopharyngeal function was categorized as a success, whereas borderline insufficient or insufficient velopharyngeal function was categorized as a failure and a secondary operation was recommended.

Complications were recorded, such as bleeding requiring transfusion, carotid arterial injury, flap dehiscence, hyponasal speech, and obstructive sleep apnea. Polysomnography was conducted if a child exhibited postoperative obstructive signs or symptoms. Possible need for a secondary operation was also documented (e.g., flap revision or take-down, dilation of the lateral pharyngeal ports, tonsillectomy, or adenoidectomy).

Statistical Analysis

Patient characteristics and descriptive statistics were summarized and the incidences of speech characteristics, and overall velopharyngeal function, were calculated. Fisher's exact test was used to compare preoperative and postoperative speech characteristics. The Wilcoxon signed rank sum test was used to compare the preoperative and postoperative weighted scores. All calculated p values were two-tailed and considered significant for values of $p < 0.05$. Statistical analyses were performed using Stata version 8 (StataCorp., College Station, Texas).

RESULTS

Patient Characteristics

Forty-nine patients with a clinical diagnosis of velocardiofacial syndrome and velopharyngeal insufficiency were evaluated between 1983 and 2009. All patients had fluorescent in situ hybridization testing for a 22q11.2 deletion, which was confirmatory in 40 patients (82 percent). Five patients with Veau type I cleft palate had palatoplasty in infancy, and three who required a pharyngeal flap were included. In summary, after exclusion of the nine patients with negative genetic testing, the two of five patients with velopharyngeal competence following primary palatoplasty, and another five patients lacking postoperative speech evaluations, 33 patients remained in the study. Clinical characteristics of these 33 patients who had a pharyngeal flap are summarized in Table 1.

The median preoperative lateral pharyngeal wall movement was 22 percent (range, 0 to 90 percent). Most patients had a large circular velopharyngeal defect (Fig. 1, *above* and *center*). The width of the pharyngeal flap was designed based on videofluoroscopic findings (Fig. 1, *below*). Most patients required a wide (34 percent) or very wide pharyngeal flap (38 percent). No patient received a narrow flap. Fourteen children (42 percent) had preoperative tonsillectomy and 10 (30 percent) also had adenoidectomy. The median follow-up was 3.8 months (range, 1.6 to 105 months).

Speech Outcomes

Preoperative and postoperative speech results are listed in Table 2. The median interval between operation and speech assessment was 3.5 months (range, 1.6 to 105 months). There was a significant improvement in the median postoperative weighted speech score as compared with preoperative values ($p < 0.001$) (Fig. 2). Normal velopharyngeal function was achieved in 16 patients and borderline sufficiency was achieved in 13 patients, translating to successful correction of velopharyngeal insufficiency in 29 of 33 patients (88 percent). An example of a patient who achieved normal velopharyngeal function is included. (**See Video, Supplemental Digital Content 1,** recorded weeks before pharyngeal flap, at the age of 4 years, demonstrating severe hypernasality. The postoperative video, taken at the age of 18 years, demonstrates normal velopharyngeal function and resonance, ***http://links.lww.com/PRS/A318***.) All four patients with persistent velopharyngeal insufficiency had construction a very wide pharyngeal flap; their preoperative lateral pharyngeal wall

Table 1. Patient Characteristics

	Value (%)
No. of patients	33
Age at surgery, yr	
Median	6.4
Range	4.4–19
Female-to-male ratio	19:14 (58:42)
Veau type I cleft palate	4 (12)
Overt submucous cleft palate	6 (18)
Occult submucous cleft palate	23 (70)
Tonsillectomy*	14 (42)
Adenoidectomy*	10 (30)
Hearing loss	8 (24)
Conductive	4 (12)
Sensorineural	4 (12)

*Tonsillectomy and adenoidectomy were performed before pharyngeal flap.

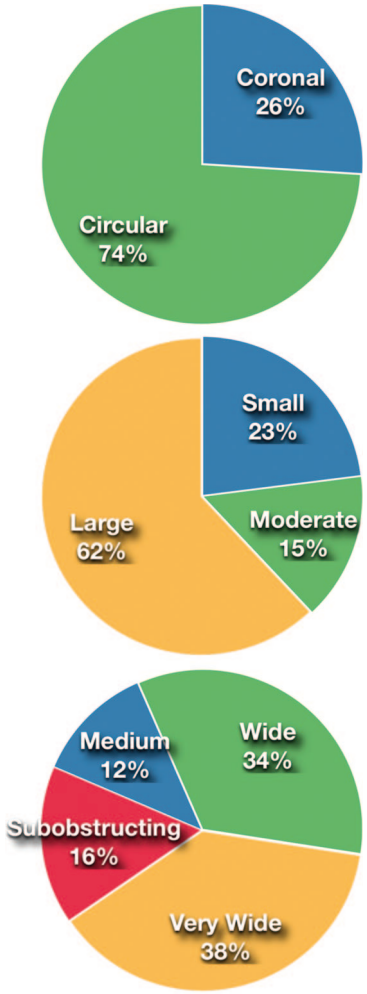


Fig. 1. Pie charts showing videofluoroscopic results for percentage of patients by (*above*) velopharyngeal closure pattern categories, (*center*) velopharyngeal gap size, and (*below*) pharyngeal flap width based on lateral pharyngeal wall motion.

movements were 5, 10, 20, and 30 percent. Two of the four patients with persistent velopharyngeal insufficiency had primary palatoplasty.

Complications

Hyponasal speech was detected in five patients (17 percent). Significant medial dislocation of the internal carotid arteries was documented in 33 percent of patients by magnetic resonance angiography (16 percent unilateral and 17 percent bilateral); an additional 21 percent had minor medial deviation of one or both carotid arteries. Injury to an internal carotid artery did not occur during any flap procedure. There was excessive intraoperative bleeding (approximately 300 cc) while elevating the flap in one patient with a medially displaced right internal carotid artery. The bleeding was controlled, but transfusion of two units of packed red blood cells was needed. One patient developed obstructive sleep apnea following a wide pharyngeal flap as confirmed by polysomnography. The patient and the family chose to treat the sleep apnea using continuous positive airway pressure therapy rather than division of the flap. One patient required augmentation of the flap. None of the patients needed division of the flap, dilatation of the lateral portals, or secondary tonsillectomy or adenoidectomy.

DISCUSSION

Children with velocardiofacial syndrome have both anatomical and physiologic palatal and pharyngeal abnormalities[33] that make operative correction of velopharyngeal insufficiency different from that for nonsyndromic children with repaired cleft palate. Learning and cognitive disabilities may also complicate speech development and therapy.[34]

Our patients generally had a large circular velopharyngeal gap with limited lateral pharyngeal wall movement. Average pharyngeal wall motion was 30 ± 26 percent in children with velocardiofacial syndrome and 58 ± 24 percent in our nonsyndromic children with velopharyngeal insufficiency after palatoplasty.[35] Velopharyngeal insufficiency can be suspected at approximately age 2.5 to 3 years. There is general agreement that children must be at least 4 years of age before velopharyngeal function can be assessed by videofluoroscopy.[36–40] The median age of videofluoroscopy in our series was 5 years. Some children ($n = 12$) with velocardiofacial syndrome and velopharyngeal insufficiency presented late at a median age of 11 years (range, 8.6 to 19 years). Patients were operated on at a median age of 6.4 years (range, 4.4 to 19 years). Six patients had an overt submucous cleft palate. Occult submucous cleft palate can be diagnosed by viewing the nasal surface of the soft palate by means of nasopharyngoscopy. Nasopharyngoscopy was performed in only two of 33 patients; nevertheless, we assumed the diagnosis of occult submucous cleft palate in all patients with a long, intact palate ($n = 23$) (i.e., no obvious signs of submucous cleft palate). Our prevalence of occult submucous cleft palate is consistent with earlier reports.[41]

Large tonsils and exuberant adenoids interfere with construction of a "tailored" pharyngeal flap. In our series, preliminary tonsillectomy was necessary in 42 percent of children and adenoidectomy was performed in 30 percent. Many authors recommend routine adenotonsillectomy be-

Table 2. Perceptual Speech Analysis*

	Pharyngeal Flap		
	Preoperatively (%)	Postoperatively (%)	*p*†
No.	33	33	
Velopharyngeal function			<0.001
Normal	0 (0)	16 (48)	
Borderline sufficient	0 (0)	13 (39)	
Borderline insufficient	0 (0)	1 (3)	
Insufficient	33 (100)	3 (9)	
Not recorded	0	0	
Resonance			<0.001
Normal	0 (0)	15 (50)	
Hyponasal	0 (0)	5 (17)	
Mixed hyponasal/hypernasal	2 (7)	3 (10)	
Inconsistent mildly hypernasal	1 (3)	3 (10)	
Moderately hypernasal	9 (30)	4 (13)	
Severely hypernasal	18 (60)	0 (0)	
Not recorded	3	0	
Nasal emission			0.005
Absent by mirror examination	0 (0)	8 (28)	
Visible	22 (76)	18 (62)	
Audible	7 (24)	3 (10)	
Not recorded	3	1	
Intraoral pressure			<0.001
Normal	1 (4)	21 (81)	
Reduced	25 (96)	5 (19)	
Not recorded	4	3	
Articulation			0.05
Normal	8 (31)	16 (62)	
Abnormal	18 (69)	10 (38)	
Not recorded	4	3	
Parental rating of speech improvement			
Slight	N/A	4 (16)	
Moderate	N/A	1 (4)	
Great	N/A	20 (80)	
Not recorded	N/A	8	

N/A, not applicable.
*Comparison of speech assessment before and after pharyngeal flap.
†*p* values calculated using Fisher's exact test.

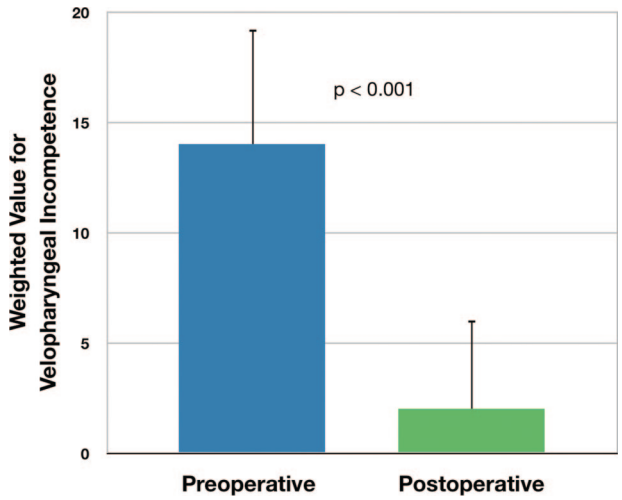
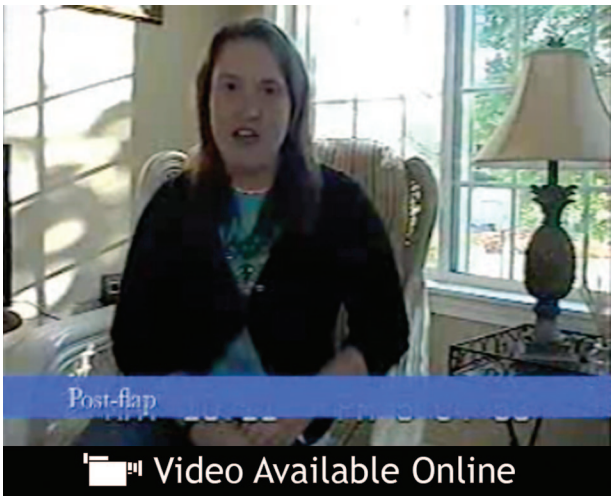


Fig. 2. Median (preoperative and postoperative) weighted values for speech symptoms associated with velopharyngeal incompetence (McWilliams and Philips, 1979). *Error bars* = SD. The *p* value was calculated using the Wilcoxon signed rank sum test.



Video. Supplemental Digital Content 1, recorded weeks before pharyngeal flap, at the age of 4 years, demonstrates severe hypernasality. The postoperative video, taken at the age of 18 years, demonstrates normal velopharyngeal function and resonance, ***http://links.lww.com/PRS/A318***.

fore a pharyngeal flap (or sphincter pharyngoplasty), regardless of size, because of postoperative hypertrophy and late-onset airway obstruction.[21,39,42,43] Our adenotonsillectomy rates might be considered low based on this advice.

There are alternative surgical options to a pharyngeal flap for correction of velopharyngeal insufficiency in children with velocardiofacial syndrome. Mehendale et al. audited their management of velopharyngeal insufficiency in 42 patients with velocardiofacial syndrome. Twenty-five patients with overt or occult submucous cleft palate were treated with radical velar muscular dissection and retropositioning.[44] Sixteen patients without obvious submucous cleft palate had augmentation of the posterior pharyngeal wall. One patient had both retropositioning and pharyngeal augmentation. Of the 25 patients managed with velar muscular retropositioning, 13 required revision, either Hynes type pharyngoplasty ($n = 11$) or a pharyngeal flap ($n = 2$). Of the 16 patients who had only pharyngoplasty, three required either revision of the pharyngoplasty ($n = 1$) or repeated velar retropositioning ($n = 2$). This report underscores the relative ineffectiveness of muscular retropositioning, as compared with pharyngoplasty. Palatoplasty reorients the velar muscles but does not address velar or lateral pharyngeal wall hypotonia or the increased velopharyngeal depth secondary to platybasia and adenoidal hypoplasia.

Sphincter pharyngoplasty only decreases the circumference of the velopharyngeal gap. Of 32 velocardiofacial patients with velopharyngeal insufficiency, Losken et al. reported a 22 percent revision rate following sphincter pharyngoplasty.[24] No description of the type of palatal anomaly was included. In a retrospective study, Witt and colleagues used sphincter pharyngoplasty in 19 patients with velocardiofacial syndrome (14 with submucous cleft palate and three with incomplete cleft of the secondary palate).[33] They found successful speech outcome in 18 of 19 patients; however, five patients developed persistent snoring and one had sleep apnea.[33] Sie et al. also documented results of sphincter pharyngoplasty in nine patients with velocardiofacial syndrome and occult submucous cleft palate. They found normal speech in five of nine patients, minor velopharyngeal insufficiency in the remaining four patients, and no patients with hyponasality.[23]

There is one report of Furlow double-opposing Z-palatoplasty in velocardiofacial syndrome as primary repair. D'Antonio et al. retrospectively reviewed four patients, and none had adequate velopharyngeal closure.[45] Details on the preoperative velar function and type of cleft palate were not reported.

Our 88 percent correction of velopharyngeal insufficiency following pharyngeal flap in velocardiofacial syndrome compares favorably with previous reports.[40,46,47] This outcome nearly equals that in nonsyndromic patients with repaired cleft palate and velopharyngeal insufficiency.[19,21,32,48] Furthermore, our success with one operation compares favorably to the rates of velopharyngeal sufficiency reported after multiple operations by Mehendale et al.[44]

Despite the effectiveness of a pharyngeal flap, many surgeons worry about possible obstructive sleep apnea. Some degree of minor obstruction, as exhibited by hyponasal speech and snoring, is desirable in the immediate postoperative period; however, this diminishes 2 to 3 months later, with reduced swelling of the flap and pharynx.[21,49–51] Shprintzen, in his initial report, found an increased rate of obstructive sleep apnea after pharyngeal flap in 28 patients with velocardiofacial syndrome (43 percent) compared with nonsyndromic cleft palate patients (6 percent).[52] In a later study from another institution, the review by Shprintzen and colleagues of a larger series of 49 patients with velocardiofacial syndrome found no obstructive sleep apnea following pharyngeal flap.[21] Only one of our patients developed airway obstruction; this same low rate was documented in nonsyndromic patients with repaired cleft palate undergoing pharyngeal flap.[35,53] Moreover, comparable rates of obstructive sleep apnea have been reported after sphincter pharyngoplasty in nonsyndromic and velocardiofacial syndrome patients.[33,54]

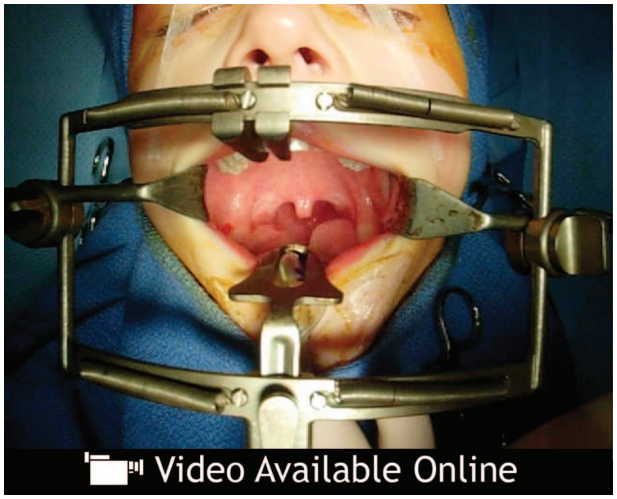
Careful preoperative analysis by the surgeon and speech pathologist is essential to planning operative correction of velopharyngeal insufficiency and minimizing sleep apnea. Despite wide variance in preoperative lateral pharyngeal wall motion (0 to 90 percent) in our series, a pharyngeal flap was successful because the width was designed based on lateral pharyngeal wall movement and the transverse pharyngeal dimension. There is also evidence that flap width does not correlate with obstructive sleep apnea.[21,42,52,55] Indeed, 72 percent of our patients had a wide or very wide pharyngeal flap, yet only one patient developed sleep apnea after a wide flap.

A second feared complication of pharyngeal flap in patients with velocardiofacial syndrome is injury to a medially transposed internal carotid artery. Tatum and colleagues used magnetic resonance angiography to show that 25 percent of

patients had medially displaced internal carotid arteries lying directly beneath the pharyngeal flap donor site.[48] Indeed, some surgeons believe that medial malposition of the internal carotid arteries is a relative contraindication to pharyngeal flap in patients with velocardiofacial syndrome.[56,57] Deviated internal carotid arteries can be seen during the procedure. (**See Video, Supplemental Digital Content 2,** which shows pulsations of medially positioned internal carotid arteries, ***http://links.lww.com/PRS/A319***.) As in other reports,[21,58] injury to the internal carotid artery did not occur in our series, 33 percent of which were medially deviated. Nevertheless, one patient with a medially deviated internal carotid artery had excessive blood loss during elevation of the flap. The source of bleeding in this patient was not entirely clear; the internal carotid artery was not injured. Others have noted that bleeding in this stage in the operation is caused by cutting the ascending pharyngeal artery or one of its branches.[28,59]

Mitnick et al. reported that visible pulsation seen on nasopharyngoscopic evaluation does not correlate with medially deviated internal carotid arteries, and that deviated internal carotid arteries seen on magnetic resonance angiography do not always exhibit obvious pulsations.[60] These authors concluded that vascular imaging should be performed on all patients with velocardiofacial syndrome before a pharyngeal flap. To the contrary, Witt and colleagues concluded that preoperative imaging is not cost effective and that there are no reports of patient deaths or internal carotid artery injury.[61] In his editorial comment, Shprintzen expressed concern over the small number of patients, the lack of cost-to-benefit analysis, and reliance on anecdotal evidence.[62] He concluded that magnetic resonance angiography serves to prepare the surgeon.[48,60] We agree, and still recommend it. Interestingly, often parents are aware of this anatomical variant in this disorder (from the Internet) and ask for an explanation as to its importance.

As in any study based on subjective speech evaluation, there are limitations. In our cleft lip–cleft palate clinic, two speech pathologists who specialize in cleft palate evaluated all the patients. Judgments may differ slightly and continuity with one speech pathologist would have been preferable.



Video. Supplemental Digital Content 2 is an intraoperative video showing pulsations of medially positioned internal carotid arteries, ***http://links.lww.com/PRS/A319***.

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PATIENT CONSENT

Parents or guardians provided written consent for the use of the patient images.

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