

Submucous cleft palate: from diagnosis to management

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Background/aim: The aim of this study was to investigate submucous cleft palate (SMCP) patients in order to document the age of diagnosis, cause of referral, symptoms and palate findings, objective evaluation of nasalance and velopharyngeal dysfunction (VPD), and intervention type.

Material and methods: The archive of Hacettepe University Cleft Lip and Palate, Craniomaxillofacial Anomalies Research, Treatment, and Application Center was reviewed retrospectively, and 166 patients diagnosed with SMCP were documented.

Results: The mean age of the patients at the time of initial referral was 10 years and 3 months \pm 8 years and 5 months with the youngest being 1 month and the oldest 44 years old. The primary complaint of 127 (76.5%) patients was speech disorder. Seventy-nine patients underwent surgery, and the mean age was determined as 10 years \pm 6 years and 8 months.

Conclusion: The diagnosis of SMCP continues to be rather late, being delayed until a prominent speech disorder is present. Studies for the awareness of health professionals are of great importance for the early diagnosis and intervention of SMCP in order to prevent adverse effects.

Key words: Submucous cleft palate, velopharyngeal dysfunction, speech disorder

1. Introduction

Submucous cleft palate (SMCP), first described by Roux in 1825, is a congenital disorder that is a relatively rare variant of cleft palate and has specific anatomic and clinical features (1). Bifid uvula, a notch in the posterior end of the hard palate, and zona pellucida in the midline of the soft palate due to muscular diastasis are the three clinical features of SMCP stated by Calnan in 1954 (2, 3). For asymptomatic SMCP, Kaplan introduced the term "occult SMCP" to describe patients who had velar malfunction but did not exhibit any of the anatomical signs of the classic triad (4). Although cleft palate is usually diagnosed easily in the newborn period during the initial screening, diagnosis of SMCP is often delayed. Depending on the presence of velopharyngeal dysfunction (VPD), patients may develop symptoms at any age or may remain asymptomatic throughout their life. Submucous cleft palate may be identified by a subjective speech assessment or objective tests such as functional magnetic resonance imaging or nasopharyngoscopy, whereas these

examinations are difficult to perform in patients under 2 years of age (5). Moreover, once diagnosed, physicians occasionally plan follow-up for the patients with regular visits for a certain period of time; therefore, SMCP tends to be treated surgically later than other types of cleft palate (6).

The aim of this study was to retrospectively review the SMCP patients treated at Hacettepe University Cleft Lip and Palate, Craniomaxillofacial Anomalies Research, Treatment, and Application Center. The content of the investigation was the age of diagnosis, gender distribution, cause of referral, symptoms and palate findings, objective evaluation of nasalance and VPD, and intervention type.

2. Materials and methods

This retrospective study, following approval of the institutional review board (GO 17/633-31), was conducted at Hacettepe University Cleft Lip and Palate, Craniomaxillofacial Anomalies Research, Treatment and Application Center; currently, the only officially approved

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center in Turkey. One hundred and sixty-six patients diagnosed (82 females (49.4%) and 84 males (50.6%)) with SMCP between January 2011 and January 2018 were included in the study. The demographic and preoperative data, including date of birth, sex, age at diagnosis, referral reason, family history of cleft, symptoms, either type of SMCP (2, 4), presence of cleft lip and accompanying syndrome/disease, age at repair, intervention type, first visit/preintervention and/or postintervention, and results of flexible nasoendoscopy assessment and nasometric evaluation, were reviewed.

2.1. Assessment protocol

Assessment protocol included speech evaluation via speech nasometer and flexible nasoendoscopic assessment both at the first visit and postoperatively with intraoral examination.

2.1.1. Intraoral examination

This exam was done by an experienced plastic surgeon, a speech language therapist, and an ear-nose-throat surgeon to assess any signs of cleft palate/SMCP or any other intraoral anomalies at the first visit.

2.1.2. Speech assessment

Instrumental assessment was done via Nasometer II Model 6450 (Kay Elemetrics, Lincoln Park, NJ, USA) to collect objective data. The data were collected at a voice-speech laboratory in a sound-treated room. Calibration was performed in the room at the start of every day before data collection. Syllables created by combining high oral pressure consonants with low /a/ and high /i/ vowels [pa/, pi/, ta/, ti/, ka/, ki/, sa/, si/, fa/, fi/, tfa/, tfi/] were repeated five times each. Running speech was assessed via counting from 1 to 10, and phrases loaded with high oral pressure consonants and /s/,/t/,/p/ phonemes in Turkish were measured (Appendix 1).

2.1.3. Flexible nasoendoscopy assessment

Visualization of the velopharyngeal port by the use of a flexible nasoendoscope during phonation was done at the Department of Ear-Nose-Throat (ENT), Head and Neck Surgery, Hacettepe University by an ENT surgeon and a speech-language therapist. Evaluation was made with an ENTity SD LED Nasoendoscope, STORZ TELECAM DX II camera (KARL STORZ SEÖ, Tuttlingen, Germany), and STORZ LED NOVA 150 light source (KARL STORZ SEÖ, Tuttlingen, Germany) with diameters of 2.2 mm for the pediatric group and 4 mm for the adult group. Velopharyngeal sphincter was assessed while the patients repeated high oral pressure consonants combined with /a/ and /i/ [pa/,pi/,ta/,ti/,ka/,ki/,sa/,si/] and kirksekiz (48; a number in Turkish) three times each and counting from 1 to 10 in Turkish. Velopharyngeal closure type and gap were determined by an experienced speech therapist. Velopharyngeal closure types were noted as coronal,

circular, sagittal, or circular with Passavant's ridge, and velopharyngeal port closure during phonation was noted as complete, minimal gap, moderate gap, or absent (7, 8). Phoneme-specific nasal emission findings were also noted.

2.2. Intervention

The type of intervention was determined depending on the patient's age, speech assessment, and flexible nasoendoscopy findings.

2.3. Statistical analysis

SPSS 22.0 (IBM Corporation, Armonk, NY, USA) software was used for statistical analysis. Distribution of the numerical variables was evaluated with the Shapiro-Wilk test, and quantitative data are presented as mean \pm standard deviation, and median (min, max), and categorical variables are presented by frequency (percentage). The variables were examined at 95% confidence level, and $P < 0.05$ was considered significant. The Kruskal-Wallis test was used to assess for significant differences of nonparametric continuous dependent variables by a categorical independent variable with three or more groups. Dunn's test was used for pairwise comparisons when the difference between the groups was significant. The Mann-Whitney U test was used to compare differences between two nonnormally distributed independent groups. For nonnormally distributed paired two-group comparison, the Wilcoxon signed rank test was used. Spearman's correlation coefficient was used to measure the strength and direction of the association between two ranked variables.

3. Results

The demographic data of the patients included in the study is summarized in Table 1. Of the 166 patients, 82 (49.4%) were female and 84 (50.6%) were male. Forty-two (25.3%) patients had syndromes or accompanying anomalies. The mean age of the patients at the time of initial referral was 10 years and 3 months \pm 8 years and 5 months with the youngest being 1 month and the oldest 44 years old. The number of patients undergoing surgery was 79, and the mean age was determined as 10 years \pm 6 years and 8 months. The age distribution of surgery timing ranged from 5 months to 39 years, with a median age of 7 years and 3 months (Table 1).

The primary complaint of 127 (76.5%) patients who applied to our center was speech disorder. Interestingly, 13 (10.23%) patients out of 127 had the complaint of initiation of speech disorder after having adenoidectomy. Seventeen patients (8.4%), who were below 3 years old, were referred to the center with the suspicion of submucous cleft palate.

Upon examination for the presence of cleft lip, nine (5.4%) individuals were found to have it; two of the clefts were bilateral, two were right- and five were left-sided.

Table 1. Demographic data and intervention types.

	N (%)
Sex	
Female	82 (49.4)
Male	84 (50.6)
Total	166 (100)
Age at surgery (years)	
<3	6 (7.59)
3–6	15 (18.98)
6–8	22 (27.84)
8–10	9 (11.39)
10–18	17 (21.51)
>18	10 (12.65)
Total	79 (100)
Intervention type	
Follow-up	88 (53)
Surgery	
Furlow palatoplasty	36 (21.7)
Pharyngeal flap	29 (17.4)
Intravelar veloplasty	7 (4.2)
Posterior pharyngeal wall augmentation	6 (3.6)
Total	166 (100)

N: number, %: percentage.

A normal uvula appearance was seen in 111 (66.87%) patients, 40 (24.09%) had bifid uvula, 10 (6.02%) had short uvula, and three (1.81%) had both bifid and short uvula. The number of patients with an intraoral velar notch during phonation was 15 (9.04%).

Both parental consanguineous marriage and family history were determined through questioning. While 137 (82.5%) did not have parental consanguineous marriage, 29 (17.5%) did. No family history of any cleft type existed for 139 (83.7%) patients; however, 27 (16.3%) had cleft history in the family. Of those 27 patients, 10 (6%) had a sibling, three (1.8%) had a mother, two (1.2%) had a father, and two (1.2%) had a child who had SMCP, and 10 (6%) had distant relatives who had cleft history.

Flexible nasoendoscopy assessment was performed on 114 (68.6%) patients at their first visit and/or after intervention. The mean age of the patients was 11 years and 7 months with a median of 8 years and 8 months. The youngest patient was 3 years and 1 month old, and the oldest was 44 years old. Patients' velopharyngeal motility functions as velopharyngeal valve closure types (Figure) and gap findings were outlined in Tables 2 and 3,

respectively. Flexible nasoendoscopic assessment was not completed on 52 (31.33%) patients because either their age was not appropriate, or they had their first intervention at another center, or they were not cooperative. Although 40 patients showed complete closure of velopharyngeal valve, 27 (67.5%) patients showed high nasalance scores ($P < 0.001$), preoperatively.

Nasometric evaluation was performed on 132 patients at their first visit. The mean age of the patients was 11 years and 9 months, with a median of 8 years and 4 months. The youngest patient was 2 years and 7 months old, and the oldest was 44 years old. Fifty-nine patients had both preoperative and postoperative nasometric assessment. The mean age of the patients was 8 years and 8 months with a median of 7 years and 5 months. The youngest patient was 2 years and 7 months and the oldest was 36 years old. Postoperative nasalance scores of syllables and sentences were statistically lower compared to preoperative scores ($P < 0.05$) (Table 4).

When the speech results of the two leading surgical options, i.e. Furlow palatoplasty and pharyngeal flap, were compared with regard to nasalance scores, the difference was not statistically significant ($P > 0.05$). As seen in Table 5, all nasalance scores, except /si, ja/ for preoperative velopharyngeal gap and /ti, ka, si, tji/ and P-loaded sentences for postoperative velopharyngeal gap, had a relationship with velopharyngeal gap. As the velopharyngeal gap increases, the score of nasalance also increases ($P < 0.05$).

The mean duration between surgery and postoperative assessment was 8 months (0.83 ± 0.13) and the median value was 4 months.

4. Discussion

The diagnosis of SMCP can often be ignored because of the wide variation in anatomical abnormalities and it is mostly not evident before the child begins connected speech. In our study, the mean age at diagnosis was 10 years and 5 months (ranging from 3 months to 44 years). Previous literature shows that only 10% of SMCP cases are symptomatic (9). In the studies by Ha et al. and Oji et al., for most of the cases, the prominent complication at the time of diagnosis was speech disorder (5, 10). Similarly, in our study, 76.5% of the patients applied to the clinic because of a speech disorder; however, 66.9% had a normal appearance of the uvula. These findings support the hidden feature of SMCP. Even for the patients who consulted a physician at least once in their life, our results showed that the age of diagnosis was rather late compared to the previous studies (1, 5, 6, 10, 11). The reasons for the very late diagnosis in this study are thought to be: 1. Either SMCP was not known to every physician or there was lack of awareness of differential diagnosis, 2. Besides

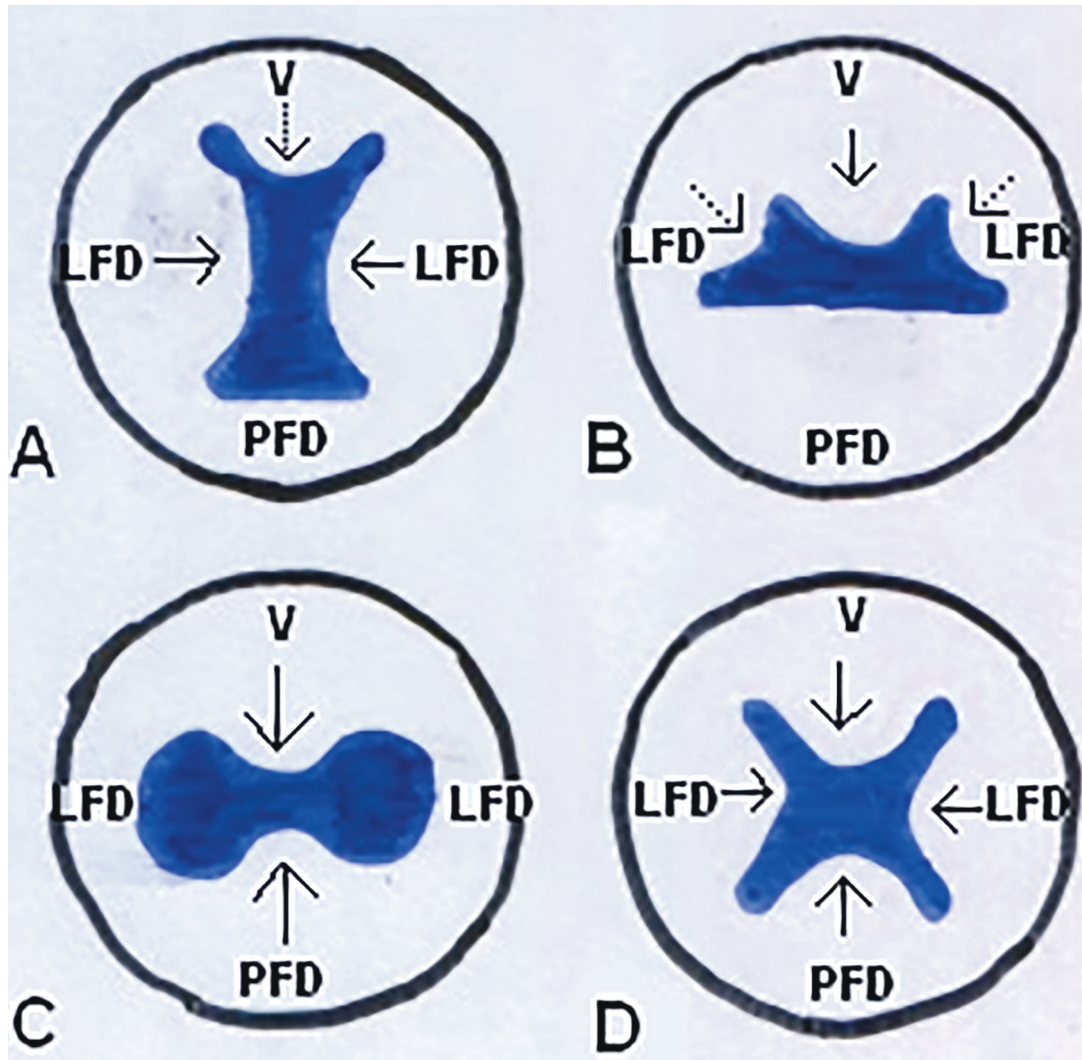


Figure. Illustration of velopharyngeal closure types. A: sagittal, B: coronal, C: circular with Passavant's ridge, D: circular (arrow: main movement, dot arrow: secondary movement, V: velum, LFD: lateral pharyngeal wall, PFD: posterior pharyngeal wall) (35).

the lack of speech–language therapists in the country, the number of experienced therapists within a cleft lip–palate and craniofacial anomalies team, which is crucial for the differential diagnosis of SCMP, was too small, 3. The families did not know where to apply.

The degree of speech defect depends on the extent of SMCP; if it is extensive, then the child's speech may not differ from that seen in cases of an overt cleft palate. Less severe SMCP may cause minimal nasality partly because of the degree of movement of the soft palate and partly because the adenoid pad could act as a compensatory factor. However, this minimal nasality can progress into a severe case following adenoidectomy (8).

Reiter et al. reported that before diagnosis of SMCP, 29% of the patients had an adenoidectomy and 12% had

a tonsillectomy. After adenoidectomy or tonsillectomy, 22% noticed a new hypernasal speech or enhancement of preexisting hypernasal speech (11). This was also described before by Saunders et al. (12). In our study, 13 patients had adenoidectomy before the diagnosis of SMCP. Although the palate is an important component of velopharyngeal valving, it is not the only one. The palate, the velum in particular, is simply one component of the velopharyngeal mechanism, and there is quite a bit of variability in the size and shape of the other structures within the pharynx. The size of the adenoid is of particular importance because the adenoid sits in the plane of velopharyngeal closure in young children. In fact, velopharyngeal closure in children up until at least 6 years of age is actually veloadenoidal. Based on some studies, adenoid size may be the most

Table 2. Velopharyngeal closing pattern.

Velopharyngeal closing pattern					
	Circular N (%)	Coronal N (%)	Circular with Passavant's ridge N (%)	Sagittal N (%)	Other (with tongue compensation) N (%)
Initial	62 (54.38)	39 (34.21)	9 (7.89)	4 (3.50)	1 (0.88)
After intervention	29 (51.78)	22 (39.28)	4 (7.14)	1 (1.78)	

N: number, %: percentage.

Table 3. Velopharyngeal gap.

Velopharyngeal Gap					
	No gap N (%)	Minimal N (%)	Moderate N (%)	Complete N (%)	PSNE N (%)
Initial	40 (35.08)	26 (22.81)	17 (14.91)	23 (20.18)	8 (7.02)
After intervention	23 (41.07)	16 (28.57)	7 (12.50)	8 (14.28)	2 (3.57)

N: number, %: percentage, PSNE: phoneme-specific nasal emission.

Table 4. Nasalance scores.

	Preoperative nasalance scores \pm SD	Postoperative nasalance scores \pm SD	P
pa	41.91 \pm 19.84	33.00 \pm 21.05	0.000*
pi	63.58 \pm 19.89	53.73 \pm 25.81	0.000*
ta	45.44 \pm 18.69	33.44 \pm 19.64	0.000*
ti	68 \pm 18.27	54.49 \pm 24.86	0.000*
ka	44.9 \pm 20.70	33.34 \pm 20.32	0.000*
ki	67.21 \pm 20.44	56.27 \pm 24.97	0.015*
sa	49.46 \pm 17.99	36.32 \pm 20.34	0.000*
si	71.33 \pm 16.74	55.83 \pm 27.12	0.000*
fa	46.62 \pm 18.05	36.32 \pm 20.32	0.005*
fi	67.59 \pm 15.48	57.66 \pm 24.70	0.082
tfa	46.87 \pm 17.67	36.22 \pm 20.15	0.002*
tfi	67.74 \pm 16.62	56.29 \pm 24.05	0.014*
Counting 1-10	57.74 \pm 18.61	50.80 \pm 20.65	0.003*
S-loaded sentence	55.51 \pm 19.57	46.27 \pm 21.17	0.027*
T-loaded sentence	56.90 \pm 18.49	48.02 \pm 21.49	0.007*
P-loaded sentence	52.87 \pm 18.05	45.66 \pm 20.55	0.045*

*P < 0.05, SD: standard deviation.

important factor in achieving velopharyngeal closure. As the adenoids are important to normal speech production, it is clear that adenoidectomy should be avoided in

individuals with cleft palate SMCP unless there is serious upper airway obstruction because of the potential for development of VPD (13). Similar to other studies (11),

Table 5. Nasalance scores in relation to preoperative and postoperative velopharyngeal gap.

Nasalance scores vs velopharyngeal gap	Preoperative P	Postoperative P
pa	0.000*	0.043*
pi	0.000*	0.036*
ta	0.000*	0.009*
ti	0.000*	0.070
ka	0.000*	0.093
ki	0.001*	0.047*
sa	0.170*	0.044*
si	0.060	0.058
fa	0.060	0.042*
fi	0.008*	0.038*
tfa	0.014*	0.046*
tfi	0.011*	0.072
Counting 1-10	0.000*	0.005*
S-loaded sentence	0.011*	0.015*
T-loaded sentence	0.001*	0.027*
P-loaded sentence	0.000*	0.087

*P < 0.05

our study showed that hypernasal speech was the primary symptom of VPD.

Findings of the intraoral examination on our patients were in accordance with those of the previous studies (5, 11). Therefore, the diagnostic criteria should not be the presence of all three factors of the triad but should require only at least one or even the absence of the typical signs if combined with nasoendoscopic findings. In this process, a bifid uvula is the first important feature for differential diagnosis of SMCP. Although the so-called occult SMCP does not exhibit this Calnan's triad sign (4), it has been reported in recent years that bifid uvula is evident with differing ratios in SMCP cases (5, 10). As in our study, 40 (24.1%) patients had bifid uvula; the classical triad does not necessarily have to be present. Thus, the diagnosis should be considered whenever one of these characteristics is found in a patient, especially if a speech disorder with nasality is also present (14, 15).

In our study, the frequency of the circular pattern (36.7%) was found to be the highest followed by a coronal pattern (23.5%), which is in accordance with Mardini et al.'s study (59.7% and 29.8%, respectively); however, it is in contrast to the findings of Garcia et al., who reported 37% and 44% incidence, respectively (14, 16). In another study, a higher incidence of a circular closure pattern (62.31%)

followed by a coronal closure pattern (33.21%) in 268 submucous clefts was reported (17). Ng et al. found 73% incidence of circular closure pattern and 17% of coronal closure pattern in both syndromic and nonsyndromic patients (18). The circular with Passavant's ridge pattern was at the third rank in all of the above-mentioned studies, and the sagittal pattern was the least frequently seen pattern. Interestingly, Ng et al. did not find any sagittal patterns (18).

Speech is assessed at the level of syllables, single words, sentences, and spontaneous speech, and the key components that are evaluated are resonance, nasal airflow, and articulation. Various scoring systems have been developed to quantify speech abnormalities; however, there is great variability between centers in the collection and analysis of data (19). As a result, although perceptual speech assessment is the primary method for diagnosing and assessing VPD, indirect objective methods of evaluation, such as nasometry, can also prove useful; especially combined use of nasometry with nasendoscopy provides objective measures of velopharyngeal function (20). Speech is one of the most important parameters to assess when determining the outcome of cleft palate repair or related velopharyngeal surgeries or speech therapy (3, 21). As in our study, quantitative data obtained before and after intervention showed statistically significant changes, so the progress of speech improvement highlighted objectively.

Furlow palatoplasty and pharyngeal flap are the most frequently used surgical techniques for the management of SMCP (7, 10, 16, 21-23). However, controversy remains regarding which procedure results in the optimal outcome. The main purpose of either surgery is to improve velopharyngeal function (20). Calis et al. reported that both Furlow palatoplasty and the pharyngeal flap procedure combined with intravelar veloplasty are effective in the treatment of SMCP; they also suggested a contribution of pharyngeal flap if the patient has significant signs of hypernasality (3). However, some authors noted the complications of the pharyngeal flap procedure (21, 24) such as developing snoring and obstructive sleep apnea. Ha et al. mentioned only the complication of snoring (10). In highlighting the complications of the pharyngeal flap procedure, Chen et al. documented the effectiveness of Furlow palatoplasty (24). In the study by Swanson et al., Furlow palatoplasty was the primary surgery. In addition to the absence of the complications of the pharyngeal flap procedure, Furlow palatoplasty has the advantage of lengthening the soft palate, a goal that is particularly useful in the mobile palate (25). Sommerlad et al. reported a similar strategy for SMCP repair, using intravelar veloplasty surgery as the first procedure (22). Similar success with Furlow palatoplasty was also reported (26). In this study,

patients' nasalance scores were statistically improved after both Furlow palatoplasty and the pharyngeal flap; however, there was no statistically significant difference. Acceptable speech depends on normal resonance and the ability to produce pressure consonants orally, both of which are highly dependent on velopharyngeal closure (13). Our results demonstrate that both surgical methods can be applied to repair SMCP.

The age range when surgery was performed was found to be between 3 and 6 years (7) and between 5 and 9 years (22). In our study, the patients had their first surgical repair between 6 and 8 years of age, mostly with a median age of 7 years and 3 months and the admission to our center at quite late ages is the reason for the delay in surgical repair. However, in our study, we observed that the age of the patient at the time of surgery did not have any statistically significant effect on improvement of nasalance scores. This finding suggests that velopharyngeal motility can be achieved regardless of the patient's age. The rationale behind early surgical correction is avoidance of compensatory speech habits and intervention at an early stage in speech development (7).

Although VPD is mainly dependent upon the velopharyngeal closure pattern and the extent of anterior displacement of the palatal muscles, meaning it can therefore present clinically with various degrees of hypernasal resonance, nasal emissions, and compensatory articulations, the velum of SMCP patients is thin regardless of the velopharyngeal valve function. As a result, there may be more transpalatal transmission of sound energy through it to the nasal cavity, and even if the velopharyngeal valve is functioning normally, there may be hypernasality due to this defect (15, 27). In support of this statement, in our study, 67.5% of the SMCP patients having complete closure of the velopharyngeal port had high nasalance scores.

Measuring the outcomes following surgical management of VPD mainly focuses on speech outcome evaluation which provides a means of direct evaluation of intervention efficacy. However, there is a lack of consensus regarding the timeline and procedures for the evaluation of speech and its outcome after intervention. A literature review reveals that speech outcome evaluation time and procedures change from 1 week to 14 months after surgery and from perceptual to objective measurements (28–33). In our study, the mean duration between surgery and postoperative assessment was 8 months (0.83 ± 0.13) and the median value was 4 months with measurement procedures being objective. Willging also recommends repeated assessment of velopharyngeal function 3 months postoperatively (34).

In conclusion, the strengths of this study are the objective data of speech outcome on the basis of nasalance and VPD and the number of patients included. Our results confirm the preliminary results indicating that SMCP continues to be diagnosed rather late, with the mean age of diagnosis being higher compared to the previous studies. As the diagnosis of SMCP is delayed until a prominent speech disorder is present, we highly recommend intraoral examination during routine follow-up of a physician and if necessary, referring to an experienced center for detailed examination for any signs of SMCP. In addition to the studies to increase awareness of SMCP among all health professionals, especially for the signs of SMCP, there should be collection of objective data in order to make differential diagnosis and intervention decision and to monitor improvement after intervention. The centers should determine a follow-up protocol for SMCP patients.

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Appendix 1.

1. Counting from 1 to 10 in Turkish

/bir, iki, ytʃ, dʒrt, beʃ; alti, jedi, sekiz, dokuz, on/ - International Phonetic Alphabet
[bir, iki, üç, dört, beş, altı, yedi, sekiz, dokuz, on]

2. High oral pressure sentences

a. T-loaded sentence

/Tarık, tatlı turta jedi/ - International Phonetic Alphabet

[Tarık, tatlı turta yedi]

b. S-loaded sentence

/Sezer, sarı sakızı aldı/- International Phonetic Alphabet

[Sezer, sarı sakızı aldı]

c. P-loaded sentence

/Polatlı'ya paket posta geldi/- International Phonetic Alphabet

[Polatlı'ya paket posta geldi]