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ORIGINAL RESEARCH - ARTICLE

Velopharyngeal Insufficiency in 22q11.2 Deletion Syndrome Patients: An Assessment of Nasal Endoscopy and Speech Evaluation Findings

Ashley Soaper, MD [1] | Lara Reichert, MD MPH [4,5] | Jennifer Arzu, MPH [3] | Benjamin Yu [2] | Margaret Mumm [3] | Laura Rosenthal, MD [3,4*]

1. University of Illinois at Chicago, Chicago, Illinois
2. Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio
3. Northwestern University, Feinberg College of Medicine, Chicago, Illinois
4. Ann and Robert H. Lurie Children's Hospital of Chicago, Chicago, Illinois
5. Albany Medical Center, Albany, New York

Abstract

Objective: The goal of this study was to assess the correlation between nasal endoscopy and speech evaluation findings in a population of 22q11.2DS patients undergoing operative intervention for correction of VPI at a quaternary care pediatric hospital. We hypothesized using a modified grading scale, certain nasal endoscopy findings such as larger pre-operative velopharyngeal gap size would correlate with perceptual speech assessment findings such as decreased intelligibility, increased hypernasality, and increased nasal emission.

Subjects and Methods: A retrospective chart review was conducted, reviewing fifty-two pediatric patients with 22q11.2DS who presented to the VPI clinic and were candidates for surgery at an urban, quaternary care children's hospital between 2010 and 2019. The following information was collected: demographic information, presenting symptoms of 22q11DS, speech therapy evaluation data, VPI surgical data, and complications. Thirty-two patients had complete pre-operative records and thus were ultimately included in the review.

Results: Twelve patients (37.5%) had a small gap size, 13 patients (40.6%) had a moderate gap size, and 5 patients (15.6%) had a large gap size. There was no statistically significant correlation between intelligibility and right lateral wall movement (rho: 0.315, p-value: 0.218), left lateral wall movement (rho: 0.175, p-value: 0.502), or AP movement (rho: 0.172, p-value: 0.525). Velum length was negatively correlated with intelligibility (rho:-0.486, p-value:0.048).

Conclusion: Our data emphasizes the importance of a multidisciplinary team when evaluating complex 22q11.2DS patients. Our unique scale provides an objective measure of nasal endoscopy by consistently documenting the findings of AP movement, gap shape, gap size, as well as other details such as velar notch and presence of Passavant's ridge.

Keywords: velopharyngeal insufficiency, 22q11.2 deletion syndrome, nasal endoscopy

1 | INTRODUCTION

22q11.2 deletion syndrome (22q11.2DS), also known as velocardiofacial syndrome (VCFS), DiGeorge syndrome, 22q11.2 related disorders, amongst others, is the most common human microdeletion syndrome, with an estimated incidence of 1:4000 births (1). 22q11.2DS presentation is highly heterogeneous with varying phenotypes, including congenital heart disease, learning impairme -nts, neonatal sepsis, seizures, and velopharyngeal insufficiency (VPI). Given the variable presentation, the diagnosis has historically been made outside of the neonatal period.

22q11.2DS is the most common syndrome associated with VPI, with an incidence ranging from 32% to 90% (2),(3). Symptoms of VPI include hypernasality, nasal air escape, and misartic -ulation, with frequent causes including hypotonia of the velopharyngeal muscles, asymmetric muscle function, or poor coordination of velopharyngeal closure (4). Some patients present with dysphagia in the form of nasal regurgitation. A team-based approach between speech language pathologists and otolaryngologists is impera -tive to care for these complex patients, as some will ultimately require corrective surgery (5),(6),(7). Surgical management most commonly includes pharyngeal flap or dynamic sphincter pharyngoplasty, amongst others.

An objective grading system for assessing VPI via nasal endoscopy was first described by Golding-Kushner in 1990 (8). The scale rates right and left lateral wall movement (0 to 0.5), right and left palate movement (0 to 1.0), and posterior wall movement (0 to 1.0) (9). In 2012, a panel suggested the intra-rater scores of this scale were overall consistent, but there was a wide range of scoring between different raters, thus showing it is not generalizable to a wide population at multiple centers (9). At our institution, we modified the Golding-Kushner scale to include velopharyngeal gap shape and size. Each examination is scored simultaneously by two otolaryngologists in conjunction with at least one speech pathologist.

Speech intelligibility, defined as the percentage of words understandable to an unfamiliar listener, is affected by a variety of factors, only one of which is resonance. Intelligibility is also dependent on the listener. Specific speech measurements have been developd to attempt to isolate and score hypernasality and nasal emission independently from other speech articulation errors. Similarly, nasal endoscopy is a measurement of velopharyngeal anatomical function independent of speech intelligibility or articulation. The purpose of nasal endoscopy is to assess velopharyngeal movement and aid surgical planning (10). It is not a tool for measuring speech perception, and to our knowledge, there have been no prior studies evaluating the relationship between nasal endoscopy findings and perceptual speech assessment measures.

VPI in 22q11.2DS patients often leads to poor speech intelligibility, but poor intelligibility is not specific to VPI and encompasses many other speech disorders(4). speech evaluation measures have been developed to assess the likelihood and severity of VPI based on clinical findings, including the Pittsburgh Weighted Speech Scale (PWSS), Cleft Audit Protocol for Speech-Augmented-Americleft Modification (CAPS-A-AM), and Structured Photographic Articulation Test II: Featuring Dudsberry (SPAT-D II). The PWSS includes five categories: nasal air emission, presence of facial grimace, nasality/resonance, phonation/voice, and articulation to determine the "nature of velopharyngeal valve". The PWSS score can estimate if the valve is competent, borderline competent, borderline incompetent, or incompetent. A higher PWSS score indicates more severe velopharyngeal dysfunction, where any score above 7 indicates an incompetent velopharyngeal valve (11). The CAPS-A-AM score is a validated measure that assesses the following parameters: speech acceptability (on a scale of 0-3 where 0 is acceptable and 3 is very unacceptable), voice, hypernasality (on a scale of 0-4 where 4 is severe), hyponasality (0-2), nasal air emission (0-2), consonant production, and cleft speech characteristics (12). The SPAT-D II test uses photographs of a dog character named Dudsberry to

Supplementary information The online version of this article (https://doi.org/10.52845/JORR/2023/4.1.2) contains supplementary material, which is available to authorized users.

Corresponding Author: *Laura Rosenthal, MD*
Ann and Robert H. Lurie Children's Hospital of Chicago Chicago, IL 60611
Email: LRosenthal@luriechildrens.org

test 59 consonants and 10 consonant blends and is scored as a raw score and percentile (13). All three of these tools help provide quantitative information regarding the severity of a patient's velopharyngeal insufficiency.

The goal of this study was to assess the correlation between nasal endoscopy and speech evaluation findings in a population of 22q11.2DS patients undergoing operative intervention for correction of VPI at a quaternary care pediatric hospital. We hypothesized using a modified grading scale, certain nasal endoscopy findings such as larger pre-operative velopharyngeal gap size would correlate with perceptual speech assessment findings such as decreased intelligibility, increased hypernasality, and increased nasal emission.

2 | MATERIALS AND METHODS

Institutional review board (IRB) approval was obtain -ed. A retrospective chart review was conducted, reviewing fifty-two pediatric patients with 22q11. 2DS who presented to the VPI clinic and were candidates for surgery at an urban, quaternary care children's hospital between 2010 and 2019. Patients who did not have complete medical records (including lack of speech records) and those whose pre-operative workup was not completed at our institution were excluded (n=20). Patients were identified using the following International Classification of Diseases, tenth revision (ICD-10) codes: Q93.81 (velocardiofac -ial syndrome or deletion of chromosome 22Q11) and D82.1 (DiGeorge syndrome). All included patients were evaluated by two fellowship-trained pediatric otolaryngologists and at least one speech language pathologist in the otolaryngology clinic for the above diagnoses. The following information was collected: demographic information, presenting symptoms of 22q11.2DS, speech therapy evaluation data, VPI surgical data, and complications. A standard speech sample was elicited from patients during nasal endoscopy when possible given age and cooperation. This information was obtained during their VPI clinic visits pre- and post-operatively. Nasal endoscopy measures were recorded routinely for each patient as in Table 1.

Table 1: *Grading scale for nasal endoscopy exams in the VPI clinic.*

Adenoid size, blocking X of choanae	0% 25% 50% 75% 90% 100%
Right lateral pharyngeal wall movement	0: No movement. 1: Minimal movement 2: Moderate movement 3: Good movement, walls do not touch in midline 4: Good movement, walls do touch in midline
Left lateral pharyngeal wall movement	As above
Passavant ridge	0: absent 1: present
Velum length	0: very short 1: mildly short 2: adequate
"V" on nasal surface of palate	0: absent 1: present
Velum movement, anteroposterior	0: No movement 1: Minimal movement 2: Moderate movement 3: Good movement but persistent gap in midline 4: Good movement and VP closure in midline
Velopharyngeal gap size	Pinpoint (1-9%) Small (10-25%) Moderate (25 - 75%) Large (>75%)
Velopharyngeal gap shape/closure pattern	Coronal Sagittal Circular Bow tie Pervasive gap due to minimal or no movement
Velopharyngeal gap location	Left Right Midline
Tonsil size	Absent 1+ 2+ 3+ 4+
Level of closure of attempted closure	Free text

Statistical Analysis

Descriptive statistics were calculated for patients' characteristics, with frequency and percentage reported for all categorical variables, while mean and standard deviation or median and interquartile range (IQR) are reported for all continuous variables, as appropriate. Spearman's rank correlation was used to examine the relationship between nasal endoscopy and speech evaluation findings. A two-sided p-value <0.05 was considered statistically significant.All analyses were conducted using statistical software, R version 4.1.0 within RStudio version 1.4.1717 (Vienna, Austria).

3 | RESULTS

Fifty-two patients with 22q11.2DS underwent VPI surgery between 2010-2019. The median age of 22q11.2DS diagnosis was 37.5 months (range: 0-115 months). Nine patients (17.3%) were diagnosed with 22q11.2DS because of VPI symptoms. One patient (1.9%) had a family history of 22q11.2DS.

Nine patients (28.1%) had a cleft palate. The median age at pre-operative speech evaluation was 66 months (range: 12-160 months), and the average age at the time of VPI surgery was 93 months/7.75 years (range: 36-242 months). The average age of post-operative evaluation was 104 months/8.67 years (range: 50-208 months). Surgical technique was determined based on the nasal endoscopy findings. Eighteen patients underwent dynamic sphincter pharyngoplasty, and 31 underwent pharyngeal flap. The remaining patients underwent facial artery musculomucosal (FAMM) flap (n=2) and pharyngeal injection (n=1). The average age at surgery in the pharyngoplasty group was 90 months (range: 36-170 months) and 95 months (range: 53-242) in the pharyngeal flap group.

Thirty-two patients had complete pre-operative records and thus were ultimately included in the review. Twelve of these patients underwent dynamic sphincter pharyngoplasty and 20 underwent pharyngeal flap. Two of these patients (6.3%) required revision surgery for persistent VPI.

On nasal endoscopy, the average lateral wall movement was 1.17 ± 0.70 on the right and 1.10 ± 0.66 on the left. Velum length was 1.60 ± 0.50. Anterior-posterior (AP) velum movement was 2.00 ± 0.91. Twelve patients (37.5%) had a small gap size, 13 patients (40.6%) had a moderate gap size, and 5 patients (15.6%) had a large gap size. Two patients (6.3%) did not have gap size recorded. 22 patients (68.8%) had coronal gap shape and 2 patients (6.2%) had a circular gap shape. The remaining 8 patients (25.0%) did not have the gap shape recorded. All patients' level of closure was at the inferior aspect of the adenoid.

The average pre-operative speech intelligibility was 60.7 ± 20.4%. The average intelligibility with a small gap size was 69.6 ± 18.4%. For a moderate gap size, the average intelligibility was 56.1 ± 19.8% and for a large gap size it was 41.7 ± 14.4% (Table 2). The average CAPS-A-AM hypernasality was 3.39 ± 0.72 and the CAPS-A-AM speech acceptability score was 2.33 ± 0.69. The CAPS-A-AM audible nasal emission score was 2 in all children except for one, where the score was 0. All children had a score of 0 for CAPS-A-AM hyponasality. All children except for two had a score of 0 for CAPS-A-AM voice, and the other two children had a score of 1. Only five children had pre-operative PWSS scores, with an average of 12.00 ± 7.00. Eleven children scored <40 on the SPAT-D-II test and the range was from <40 to 86 (<1 to 18th percentiles).

Table 3 demonstrates the statistical correlations between pre-operative nasal endoscopy and speech evaluation. There was no statistically significant correlation between intelligibility and right lateral wall movement (rho: 0.315, p-value: 0.218), left lateral wall movement (rho: 0.175, p-value: 0.502), or AP movement (rho: 0.172, p-value: 0.525). Velum length was negatively correlated with intelligibility (rho: -0.486, p-value: 0.048). When evaluating pre-operative CAPS-AM hypernasality score to nasal endoscopy findings, the right and left lateral wall movements have a rho: 0.074 and p-value: 0.784. The correlation between velum length and CAPS-AM hypernasality is -0.115 (p-value: 0.649). The correlation between AP movement and CAPS-AM hypernasality is -0.174 (p-value: 0.519). Gap size and CAPS-AM hypernasality have a non-statistically significant correlation (rho value: 0.412, p-value: 0.113). When evaluating the CAPS-AM speech acceptability scores, no statistically significant correlations were detected. Right and left lateral wall movement and CAPS-AM speech acceptability show a rho of 0.103 (p-value: 0.751). With velum length and CAPS-AM speech acceptability, the rho value is -0.242 (p-value: 0.425). For AP movement and CAPS-AM speech acceptability, the rho is 0.097 (p value: 0.764). Lastly, the correlation between gap size and CAPS-AM speech acceptability is 0.119 (p-value: 0.713) (Table 3).

Table 2: *Pre-operative intelligibility and gap size.*

	Small Gap	Moderate Gap	Large Gap
Pre-Op Intelligibilty n	12	9	3
Pre-Op Intelligibility Average	69.6%	56.1%	41.7%
Pre-Op Intelligbility Standard Deviation	18.4%	19.8%	14.4%

Table 3: *Pre-operative nasal endoscopy and pre-operative speech evaluation scores.*

	Pre-Op Intelligibility	Pre-Op PWSS	Pre-Op SPAT-D Score	Pre-Op CAPS-A Hypernasality	Pre-Op CAPS-A Acceptability
Pre-Op R Lateral Wall n	17		11	16	12
Pre-Op R Lateral Wall rho	0.315		-0.463	0.074	0.103
Pre-Op R Lateral Wall p-value	0.218		0.151	0.784	0.751
Pre-Op L Lateral Wall n	17	5	11	16	12
Pre-Op L Lateral Wall rho	0.175	0.000	-0.463	0.074	0.103
Pre-Op L Lateral Wall p-value	0.502	>0.999	0.151	0.784	0.751
Pre-Op Velum Length n	17	5	12	18	13
Pre-Op Velum Length rho	-0.486	0.544	0.000	-0.115	-0.242
Pre-Op Velum Length p-value	0.048	0.343	>0.999	0.649	0.425
Pre-Op AP Movement n	16	5	11	16	12
Pre-Op AP Movement rho	0.172	0.544	-0.027	-0.174	-0.097
Pre-Op AP Movement p-value	0.526	0.343	0.937	0.519	0.764
Pre-Op Gap Size n	17		11	16	12
Pre-Op Gap Size rho	-0.41		0.184	0.412	0.119
Pre-Op Gap Size p-value	0.102		0.589	0.113	0.713

4 | DISCUSSION

Although we have found some correlation between certain nasal endoscopy findings and speech scores, there lacks a strong and reproducible connection. This emphasizes the importance of a multidisciplinary team when evaluating these complex 22q11.2DS patients and that the surgical plan should be carefully tailored to each patient. It also supports the use of CAPS-AM as a useful tool for assessing velopharyngeal insufficiency. Our unique scale was developed to provide an objective measure of nasal endoscopy by consistently documenting the findings of AP movement, gap shape, gap size, as well as other details such as velar notch and presence of Passavant's ridge. However, we are still limited by inter-user variability, as there is some subjectivity in our scale. Prior investigation has supported the use of nasal endoscopy to evaluate gap size, although other tools may offer additional perspectives, such as multi-view videofluoroscopy or magnetic resonance imaging (MRI) (14).

Given 22q11.2DS patients often have stiff lateral pharyngeal walls with minimal movement, it is not data shows no significant correlation between right and left lateral wall movement and any speech score. Seemingly, the most significant correlation is between intelligibility and velum length (rho: -0.486, p-value: 0.048). Interestingly, the rho value demonstrates an inverse relationship between intelligibility and velum length in our patient cohort, which is not expected. Over half (n=17) of our cohort was noted to have an adequate velum length and 12 had a mildly short velum, so it is likely that this rho correlation is skewed given the small range of velar lengths noted. No patients in our cohort had a velum length score of 0, meaning a very short velum. Typically, children with 22q11.2DS will have a normal velum length but deep and wide oropharynx leading to poor speech. Nine patients (28.1%) evaluated had a cleft palate as well. Intelligibility is, however, not a good measure of velopharyngeal function. Rather speech acceptabi -lity on the CAPS-Am is a more appropriate measure.

There is an inverse relationship between the size of the velopharyngeal gap and intelligibility (rho: -0.41, p-value: 0.102) which is expected, as a larger gap allows for increased air escape, leading to hypernasality.

It is also expected that components of the CAPS-A-AM would have the highest association with velopharyngeal gap size as the CAPS-A-AM is our best speech perceptual measurement of hypernasal resonance(12). The right and left lateral wall movement as well as velum length show a moderate correlation with the rho value. The CAPS-A-AM aggregate score and velum length show an inverse, moderate correlation, demonstrating that as velar length gets shorter, there is more hypernasality noted.

The PWSS was designed to assess the "nature of the velopharyngeal valve" and determine its competency (11). There was a correlation between velum length and PWSS (rho: 0.544; p-value: 0.343), as well as the AP movement and PWSS (rho: 0.544; p-value: 0.343), although not statistically significant.

Many of our parameters did not show statistical significance. However, it is unknown if these findings are a result of insufficient power due to our small sample size versus truly no difference from null. Further investigation with a larger cohort of patients would be prudent to further evaluate this.

Given that our institution cares for a large 22q11.2DS population, we were interested in investigating velopharyngeal insufficiency in these specific patients. However, we intend to further investigate all non-syndromic VPI patients to see how they compare to our 22q11.2DS population. We plan to evaluate post-operative nasal endoscopy findings and speech evaluation to obtain further prognostic information for these children. Additionally, future investigation of nasal endoscopy as a tool for measuring gap size will include objective measurements of gap size from recorded images. Future investigation will also include metrics between different surgical methods (dynamic sphincter pharyngoplasty vs. pharyngeal flap) to determine whether there are any predictable differences in speech outcomes between the two techniques.

5 | CONCLUSION

Our data emphasizes the importance of a multidisciplinary team when evaluating complex 22q11.2DS patients. Our unique scale provides an objective measure of nasal endoscopy by consistently documenting the findings of AP movement, gap shape, gap size, as well as other details such as velar notch and presence of Passavant's ridge. Our data also supports the use of CAPS-AM as a useful tool for assessing velopharyngeal insufficiency and has shown some correlation between intelligibility and size of the velopharyngeal gap. Further investigation will include metrics between different surgical methods and objectively measuring velopharyneal gap sizes based off of our endoscopy images.

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