The Effect of Loudness Variation on Velopharyngeal Function in Children with 22q11.2 Deletion Syndrome: A Pilot Study

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Abstract

Objective: Children with 22q11.2 deletion syndrome (22qDS) often require surgical intervention to treat velopharyngeal dysfunction (VPD). Although some studies have documented improved velopharyngeal (VP) closure under increased speaking effort, currently no studies have examined the effect of similar behavioral speech modifications on VP closure in children with 22qDS. The purpose of this pilot study was to explore the effect of loudness on VP closure during speech in children with 22qDS and persisting VPD. Patients and Methods: Four children with 22qDS, posterior pharyngeal flap, and persisting mild VPD underwent pressure-flow testing while repeating words at habitual and increased loudness levels. Using a single-subject A-B design, descriptive statistics and graphical measures were used to examine differences in VP orifice area (VPA) and timing of closure in the habitual versus loud condition. Results: Results were mixed. Median VPA decreased during some stimuli for 3 participants, but increased for 1 subject when speaking louder. Median duration of nasal airflow decreased for 3 participants in the loud condition. Conclusion: This study presents preliminary aerodynamic data regarding the plasticity of VP physiology in the 22qDS group. Further research is needed to determine how loudness impacts VP function in children with 22qDS.

Key Words
22q11.2 deletion syndrome · Velopharyngeal dysfunction · Hypernasality · Resonance

Introduction

The velopharyngeal (VP) valve is a complex structure that serves to separate the oral and nasal cavities during speech [1]. VP dysfunction (VPD) is characterized as the inability to open and close this valve accurately and efficiently during speech. VPD can result from structural causes (e.g., insufficient palate length after palate repair surgery), neurological incompetence (e.g., dysarthria), and/or articulatory mislearning [2–5]. Children with VPD can develop compensatory articulation errors (e.g., glottal stop substitutions), nasal air emission, and hypernasal speech, which can place them at an increased risk for communication limitations and decreased social acceptance [6].

22q11.2 deletion syndrome (22qDS) is a genetic disorder characterized by palatal anomalies, facial dysmor-
The majority of children with 22qDS will have craniofacial anomalies such as submucosal cleft palate, palatopharyngeal hypotonia, VPD, and other speech and language disorders [4, 8–11]. Neuromotor differences, such as motor speech delays and cranial nerve dysfunction, have also been identified as contributing factors to VPD in children with 22qDS [2, 11–13]. Aerodynamic (pressure-flow) testing has been shown to be a valid and reliable method to assess VP closure for speech and provides indirect measures of VP orifice area (VPA) and VP closure timing [14, 15]. Previous studies using aerodynamic testing have found VPA and closure timing differences between normal speakers and speakers with cleft palate/VPD. Hypernasality can be associated with VPAs >5 mm² on pressure consonants as well as with ‘slower’ VP closure, the latter as evidenced by an increased duration of nasal airflow during speech [14–18]. Baylis et al. [2] found that children with 22qDS exhibit variability in VPA and increased nasal flow pulse durations during speech, which predict listener perceptions of hypernasality. While VP surgery is known to reduce the size of the VP orifice, the effect of behavioral speech modifications on VPA and/or VP timing in children with 22qDS remains unknown.

Surgeries such as sphincter pharyngoplasty and posterior pharyngeal flap (PPF) are considered the primary surgical treatment approaches for VPD and hypernasality in 22qDS [1, 19]. Children with this syndrome consistently exhibit hypernasality and articulation problems that are more severe than those of nonsyndromic children with VPD or cleft palate, and they do not demonstrate as favorable postsurgical speech outcomes [2, 3, 20–22]. More research is needed to determine if other behavioral treatment approaches can improve the speech of patients with 22qDS.

Increasing loudness has been posited as a speech modification that can trigger global motor activation across the speech production system and extend beyond the vocal tract to increase articulatory precision and possibility minimize VPD [23–28]. McHenry [29] examined VP closure in 28 adult subjects with VPD post traumatic brain injury by asking them to repeat the stimulus /pi/ louder. Results showed that when compared to habitual speaking levels, 89% of participants achieved a smaller average VPA on /p/ when using increased vocal effort. That study hypothesized that adults with dysarthria may be able to use louder speech as a behavioral compensation for VPD and possibly reduce hypernasality. Additional studies involving children and adults with neuromuscular deficits similar to those found in the 22qDS group have shown similar results, with louder speech correlating somewhat with improved judgments of nasality and greater articulatory excursion and precision [26, 28, 30].

If stimulated loudness produces consistent changes in VP closure, children with 22qDS and VPD may be able to benefit from use of this strategy. Those with mild VPD post secondary speech surgery may be appropriate candidates for such a trial due to already having improved VP structures (i.e., reduced VPA) as well as the desire to avoid additional revision surgeries and their associated airway risks.

The purpose of this pilot study was to examine the effect of increased loudness on VP function in a small group of children with 22qDS with prior PPF and persisting mild VPD symptoms. Specifically, the research questions of this study included: Does louder speech result in (1) a smaller VPA, and/or (2) a shorter duration of the nasal airflow pulse (i.e., duration the VP port is left open) in selected speech stimuli produced by children with 22qDS and VPD?

Method

Participants

This study was conducted at a large pediatric hospital in the Midwest and was approved by the IRB of the participating institution. Informed consent (and assent, when applicable) was obtained. Four children completed the study (3 females, 1 male, age range 6.9–14.9 years; table 1). All participants had a confirmed 22q11.2 deletion and demonstrated symptoms of mild VPD as determined through a clinical speech evaluation with a craniofacial-trained speech pathologist and plastic surgeon, pressure-flow testing, and nasopharyngoscopy. Perceptual speech and VP closure ratings during flexible nasopharyngoscopy followed recommended standardized protocols and were obtained during the participants’ routine craniofacial clinic visits and/or initial research visit [31, 32]. Mild VPD was defined as displaying mild or moderate hypernasality, audible nasal air emission and/or nasal turbulence, as well as ‘adequate’ or ‘borderline’ VPAs based on pressure-flow testing during production of the word hamper, referencing established VPA categories [14, 17].

Prior to participating in the study, all participants had undergone recent nasopharyngoscopy to confirm that no greater than ‘small’ VPAs (bilaterally, due to PPF) were present, as rated during maximum palatal excursion during speech [32]. No participant had a history of overt cleft palate, and all had previously undergone PPF surgery for VPD. When tested, time since surgery was >1 year for all participants. All spoke English as their primary language. Some participants exhibited mild developmental articulation errors such as /l/ distortions, but all had accurate articulation for oral stop consonants required for testing at the time of study participation. All participants had a history of language delay/disorder based on chart review. None had a history of permanent hearing loss or mental retardation, although some had mild learning difficulties. Most participants were informally judged to have adequate nasal patency at the time of study, although participant 2 had a history of mild fluctuating congestion.
Procedures
All participants completed pressure-flow testing during speech (PERCI-SARS™, Microtronics) [14]. By measuring differential oral-nasal pressure and volume (rate) of nasal flow, the hydrokinetic equation was used to compute estimated VPA during speech. The PERCI-SAR system was calibrated weekly throughout the testing period. Two differential pressure transducers were used to perform the pressure-flow measurements. The pressure drop across the VP orifice was measured by placing one catheter in the mouth and one in the nostril. Nasal airflow was measured through a pneumotachograph attached to clear plastic tubing that was held just inside the participant’s other nostril. Figure 1 depicts a sample computer display of the oral pressure and nasal flow peaks seen during the production of *hamper* in a normal speaker. A microphone attached to the PERCI-SARS™ hardware, approximately 12 inches from the participant’s mouth, recorded each speech sample. A digital sound level meter was placed approximately 12 inches (30.5 cm) in front of the participant’s mouth to record the average loudness level for each stimulus.

Participants were asked to repeat *pi*, *pa*, *hamper*, and *I have a hamper*, after a model. These stimuli were selected since they require complete VP closure as well as contain the /mp/ sound combination that allows for analysis of the timing of VP orifice closure (measured as the duration of the nasal airflow pulse) in a more complex speech context thought to mimic the demands of ‘conversational’ speech [14, 17]. Each stimulus was repeated five times on a continuous breath when possible, and this process was repeated two times in the habitual speech condition (HAB), given no cues. Participants were then told that they needed to produce the same set of words again, but ‘louder’. Models of ‘louder’ speech were given, with additional instruction to not yell or scream. Procedures were repeated in the louder (LOUD) speech condition. SPL (dB) was measured and recorded for each string of stimuli. A LOUD production was defined as being at least an average of 5 dB SPL greater than what that participant had produced during the HAB condition for that stimulus. If participants’ productions did not meet that level, cues were given to ‘be louder’ until the 5 dB SPL threshold (average for the utterance) was met.

Data Analysis
For each participant, the independent variable was the speaking condition (HAB vs. LOUD). The dependent variables included (1) VPA and (2) timing of VP closure as measured by duration of the nasal airflow pulse during the /mp/ sequence of *hamper* (fig. 1).

Results
VPA results for all participants are displayed in figure 2 and table 2. For participants 1, 2, and 4, median VPA decreased in the loud condition for two stimuli, but either increased or remained unchanged for the other two stim-
uli. Decreases in median VPA ranged from 0.10 to 6.0 mm². Participant 3 demonstrated a larger median VPA across all stimuli when speaking louder. For participants 1, 2, and 4, their largest VPA occurred in the HAB condition. Three of the 4 participants showed a shorter median duration of VP closure in the LOUD condition, as evidenced by decreased nasal airflow pulse duration. Decreases were by 60 ms (participant 1; HAB 245 ms; LOUD 185 ms), 45 ms (participant 3; HAB 215 ms; LOUD 170 ms), and 5 ms (participant 4; HAB 235 ms; LOUD 230 ms). Participant 2 showed no changes in median VP closure timing (HAB 280 ms; LOUD 280 ms).

### Discussion

Almost all participants showed some improvement in the extent of VP closure when speaking louder, depending on the stimulus. Visual analysis of figure 2 reveals that the maximum overall VPA for participants 1, 2, and 4 either approached or exceeded the VPA size traditionally used to define the ‘cutoff’ between adequate and inadequate closure (20 mm²) in the HAB condition [15]. Participant 3 was the only subject whose median VPA for each stimulus and largest overall VPA increased in the LOUD condition. These observations may be explained by possible underlying neuromuscular and/or VP anatomical differences at baseline, as she was the only testing subject to have a history of diagnosed hypotonia as well as the highest baseline (more severe) perceptual rating of hypernasality. These data could also suggest that increasing loudness might not facilitate improvements in the degree of VP closure for subjects with greater than borderline-mild VPD.

The majority of participants evidenced improved timing of VP closure on hamper as displayed through a reduced median duration of nasal airflow in the LOUD condition. One explanation for this finding is that changes in speech rate may have influenced timing results. A slower rate of speech has been associated with increasing loudness, and consequently improved speech clarity in normal speakers and individuals with motor speech disorders [33]. Two participants though (1, 3) increased their average speech rate when speaking louder. Decreased duration of nasal airflow was not observed in conjunction with a smaller VP gap, as median VPA increased for all subjects on hamper when speaking louder. As rate was not formally controlled in this study, the relationship between loudness, speaking rate changes, and VP function measures cannot be well explicated at this time and warrants inclusion in future iterations of this study.

The variability in VPA and duration of nasal airflow across all participants is to be expected and aligns with research regarding variation in speech motor control. When observing individual motor speech components, there can be substantial variability as different speakers strive to perform similar articulatory targets [34, 35]. The participants in the present study displayed similar variability in that mild VPD and hypernasality did not translate into similar measurements across participants of the extent and timing of VP closure in either condition. The participants’ aerodynamic profiles are distinct and support previous studies that highlight the complex relationship between VP and timing measures in the cleft palate population and 22qDS group [2, 15, 18]. It may not be possible to categorize a child with 22qDS as having one degree of VP during speech at all times, as degree of VPD may change based on the demands of the speech stimulus length and/or complexity.

An unexpected finding of this study was the variability of VPAs found in the HAB condition within each participant for each stimulus. The maximum VPAs in conjunction with an unpredictable baseline trend may also be viewed as outliers that do not reflect ‘typical’ functioning of the VP mechanism. Further statistical analysis with a greater number of participants and VPA measurements

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**Table 2. Median VPAs on /p/ (mm²) and average speech rate for hamper (syllables/s)**

<table>
<thead>
<tr>
<th>Participant</th>
<th>Pi HAB</th>
<th>Pa HAB</th>
<th>Pa LOUD</th>
<th>Hamper HAB</th>
<th>Hamper LOUD</th>
<th>I have HAB</th>
<th>I have LOUD</th>
<th>Rate HAB</th>
<th>Rate LOUD</th>
</tr>
</thead>
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<tr>
<td>1</td>
<td>0.1</td>
<td>0.2</td>
<td>0.1</td>
<td>1.5</td>
<td>2</td>
<td>8.9</td>
<td>3.1</td>
<td>2.81</td>
<td>3.14</td>
</tr>
<tr>
<td>2</td>
<td>9.6</td>
<td>9.1</td>
<td>10.7</td>
<td>5.6</td>
<td>16.5</td>
<td>4.9</td>
<td>2.8</td>
<td>2.65</td>
<td>2.60</td>
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<tr>
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<td>9.1</td>
<td>12.5</td>
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<td>16.5</td>
<td>12</td>
<td>13.5</td>
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<td>2.96</td>
</tr>
<tr>
<td>4</td>
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<td>1.6</td>
<td>0.6</td>
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<td>4.8</td>
<td>3.4</td>
<td>5.2</td>
<td>2.87</td>
<td>2.82</td>
</tr>
</tbody>
</table>
Fig. 2. VPAs on /p/ for all subjects in HAB and LOUD conditions.
is needed in order to draw more substantive conclusions of causality trends between loudness variation and VPA.

The main limitations of this pilot study involve the small sample size and relatively small number of data points for each subject across conditions. These features of the investigation limit the ability to determine statistical significance and generalize findings to a larger group of children with 22qDS. The dependent variables in this study were also strictly physiological measures associated with VP closure, whereas hypernasality is a concept that is perceptual in nature and influenced by a dynamic interplay of anatomy and physiology, as well as articulation, rate, voice, and other factors. No perceptual ratings of the HAB and LOUD productions were included in the testing protocol; therefore, to what extent a listener would have determined the participants’ hypernasality to improve is not within the scope of this study’s conclusions, nor was our intent for this preliminary study. Finally, although including participants with PPFs offers an opportunity to evaluate the effects of increased loudness as a supplement to surgical intervention, VP measurements for each child were calculated as a sum of two ports and could not account for any differences due to asymmetry in VPA and/or timing of VP closure that may be present.

This pilot study is the first behavioral investigation of VP function during speech in the 22qDS pediatric population. Results reveal that all participants tolerated testing procedures well, and cued speech changes are feasible in a pressure-flow research protocol. Future iterations of this study should incorporate a larger sample size, children who have not yet undergone VP surgery, as well as a comparison/control group. Adding a withdrawal phase in the testing protocol (i.e., A-B-A) would also provide more opportunity to establish clear trends across changes from the HAB to experimental conditions. High-quality audio recordings should be obtained so that so that blinded listener ratings of nasality for each participant could be included to allow for an analysis of the effect of louder speech on the perception of VPD symptoms. With older participants, future extensions of this study could also investigate the potential for louder speech to be used in conjunction with biofeedback, such as during nasometry and/or nasopharyngoscopy, to elicit greater closure of the VP port in a more direct ‘learning’ environment for the speaker. In sum, this study provides an important first step to consider external behavioral influences on VP closure in children with 22qDS and has set the foundation for future speech treatment research.

References


