Surgical Outcomes for the Treatment of Velopharyngeal Insufficiency in 22q11.2 Deletion Syndrome

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Abstract

This study aimed to compare outcomes of concomitant palatoplasty and sphincter pharyngoplasty with pharyngeal flap and sphincter pharyngoplasty alone for the treatment of velopharyngeal insufficiency in patients with 22q11.2 deletion syndrome. Thirty-one cases were identified for inclusion in the study. Patients were separated into 3 surgical groups: combined palatoplasty and sphincter pharyngoplasty (n = 11), pharyngeal flap (n = 7), and sphincter pharyngoplasty (n = 13). Outcome measures included perceptual speech analyses, surgical complications, and revision rates. There were no differences in preoperative speech analysis scores (P = .31). The combined palatoplasty and sphincter pharyngoplasty procedure had similar speech outcomes compared to pharyngeal flap, and both were significantly better than sphincter pharyngoplasty alone. Complication rates (P = .61) and the need for revision surgery (P = .25) were similar among all 3 groups. Concomitant palatoplasty and sphincter pharyngoplasty may be an alternative treatment for velopharyngeal insufficiency in children with 22q11.2 deletion syndrome.

Keywords

22q11.2 deletion syndrome, DiGeorge syndrome, velocardiofacial syndrome, velopharyngeal insufficiency, Furlow palatoplasty, sphincter pharyngoplasty, hypernasality, speech outcomes

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22q11.2 deletion syndrome (22qDS) has a highly variable phenotype with more than 180 associated features, including palate abnormalities and velopharyngeal insufficiency (VPI).1-8 Despite the variety of surgical techniques used to treat VPI, children with 22qDS tend to achieve inferior speech outcomes compared to non-syndromic children.9-12 Many consider the pharyngeal flap to be the most effective treatment for VPI in patients with 22qDS; however, complications such as persistent VPI, bleeding, and obstructive sleep apnea (OSA) are all well described.2,12,13

Bohm et al14 previously published that the novel combination of Furlow palatoplasty and sphincter pharyngoplasty (CPSP) was an effective treatment for VPI with superior speech outcomes and lower revision rates when compared with sphincter pharyngoplasty (SP) or pharyngeal flap (PF), respectively. However, that study excluded patients with 22qDS due to their unique anatomical differences.15-18 The primary aim of the present study is to compare surgical outcomes specifically in patients with 22qDS who underwent PF, SP, or CPSP using previously described surgical techniques.14

Methods

After obtaining institutional review board approval from Children’s Minnesota, a retrospective chart review was performed on all pediatric patients with 22qDS who underwent surgical treatment for VPI from January 1, 2008 to December 31, 2016. Thirty-one cases were identified for inclusion in the study. Patients were divided into 3 groups based on surgical intervention and were included more than once if a revision surgery was performed. Demographic data, medical history, and outcome measures were collected, including perceptual speech analyses, surgical complication rates, and revision rates.

Specialized speech pathologists performed annual perceptual speech analyses. The severity of resonance pathology was graded according to the following scale: 0 = within acceptable limits, 1 = mild hypernasality, 2 = mild to
moderate hypernasality, 3 = moderate hypernasality, 4 = moderate to severe hypernasality, and 5 = severe hypernasality. Hyponasal and mixed resonance were assigned scores of 0 and 2, respectively.

A $\chi^2$ test or Fisher exact test was used to evaluate categorical variables as appropriate for the corresponding sample size. The $t$ test and 1-way analysis of variance (ANOVA) were used to evaluate continuous variables with post hoc Tukey’s test.

**Results**

In this study, there were 11 (35%) CPSP, 7 (23%) PF, and 13 (42%) SP cases. Patient characteristics are presented in Table 1. Fifteen patients had palatal clefting pathology (48.4%). Seven patients (22.6%) underwent prior cleft repair surgery and 8 patients (26.7%) underwent prior speech surgery.

The preoperative and postoperative resonance severity scores are presented in Figure 1. The mean ± standard deviation (SD) preoperative resonance severity score for all patients was $2.9 \pm 1.5$, and there were no significant differences between the 3 groups ($P = .31$) (Figure 1). The mean ± SD postoperative resonance severity scores for the CPSP, PF, and SP groups were $0.2 \pm 0.5$, $0.3 \pm 0.5$, and $1.5 \pm 1.6$, respectively. The CPSP and PF groups both had a lower mean postoperative resonance severity score than the SP group ($P = .02$). Patients in the CPSP and PF groups had a greater improvement in resonance severity scores when compared to the SP group ($P = .006$ and .04, respectively).

The postoperative complications in the studied patient population included OSA (n = 4) and persistent VPI (n = 5) (Table 2). The complication rates for CPSP, PF, and SP were 27.3%, 14.3%, and 38.5%, respectively. There were no significant differences in the complication rates among the 3 surgical groups ($P = .61$).
A revision surgery was indicated in patients with persistent VPI and polysomnographic evidence of OSA. Six patients (19.4%) underwent surgical revision: 2 (18.2%) from the CPSP group and 4 (30.8%) from the SP group (Table 2). None of the PF patients underwent surgical revision; however, 1 required long-term continuous positive airway pressure for OSA. There were no significant differences in the surgical revision rates among the 3 groups ($P = .25$).

**Discussion**

Patients with 22qDS often have speech delays and learning difficulties, so correcting VPI in these patients is critically important. To optimize the chance of successful VPI surgery, we examined speech outcomes, complication rates, and revision rates following 3 surgical techniques. We found that patients with 22qDS who underwent either PF or CPSP had improved speech outcomes when compared to those who underwent SP.

There are several limitations to this study. First, this was a retrospective review, and consequently, the surgical intervention that patients received was not randomized, resulting in an inherent selection bias. Although not statistically significant, it should be noted that patients in the PF group had all types of clefting pathology, whereas patients in the CPSP and SP groups only had submucous or soft palate clefts. Furthermore, none of the patients in the CPSP group underwent prior cleft or speech surgery. Scar tissue that forms following a palate repair makes revision palatoplasty more technically challenging and possibly unnecessary if the levator veli palatini muscle was already realigned and the palate appropriately lengthened. Since the groups differed preoperatively in terms of their clefting type and surgical history, it is possible this biased the results. Finally, this study has a small sample size ($n = 31$) and therefore is underpowered to determine superiority of any of the 3 techniques.

**Conclusion**

CPSP may be an alternative option for the surgical management of VPI in children with 22qDS. In this preliminary study, CPSP had similar postoperative speech outcomes compared to PF and improved outcomes compared to SP alone; however, larger prospective studies are needed to validate these findings.

**Author Contributions**

Lauren A. Bohm, conception and design, acquisition analysis and/or interpretation of data, drafting/revising the manuscript, final approval, agreement to be accountable; Jessa E. Miller, acquisition analysis and/or interpretation of data, drafting/revising the manuscript, final approval, agreement to be accountable; Noëlle Morrell, acquisition analysis and/or interpretation of data, revision of the manuscript, final approval, agreement to be accountable; James D. Sidman, conception and design, acquisition analysis and/or interpretation of data, revision of the manuscript; Brianne B. Roby, conception and design, revision of the manuscript, final approval, agreement to be accountable.

**Disclosures**

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**References**


**Table 2. Postoperative Complications and the Need for Surgical Revision.**

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>All Patients (n = 31)</th>
<th>Palatoplasty + Sphincter Pharyngoplasty (n = 11)</th>
<th>Pharyngeal Flap (n = 7)</th>
<th>Sphincter Pharyngoplasty (n = 13)</th>
<th>P Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Complication</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>OSA</td>
<td>4 (12.9)</td>
<td>2 (18.2)</td>
<td>1 (14.3)</td>
<td>1 (7.7)</td>
<td>.61</td>
</tr>
<tr>
<td>Persistent VPI</td>
<td>5 (16.1)</td>
<td>1 (9.1)</td>
<td>0 (0)</td>
<td>4 (30.8)</td>
<td>.25</td>
</tr>
<tr>
<td>Total</td>
<td>9 (29.0)</td>
<td>3 (27.3)</td>
<td>1 (14.3)</td>
<td>5 (38.5)</td>
<td></td>
</tr>
<tr>
<td>Need for revision</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>6 (19.4)</td>
<td>2 (18.2)</td>
<td>0 (0)</td>
<td>4 (30.8)</td>
<td>.25</td>
</tr>
<tr>
<td>No</td>
<td>25 (80.6)</td>
<td>9 (81.8)</td>
<td>7 (100)</td>
<td>9 (69.2)</td>
<td></td>
</tr>
</tbody>
</table>

Abbreviations: OSA, obstructive sleep apnea; VPI, velopharyngeal insufficiency.

*Values are presented as number (%).


