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Factors affecting articulation skills in children with velocardiofacial syndrome and children with cleft palate or velopharyngeal dysfunction: A preliminary report

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Abstract

Objective—To examine the influence of speech perception, cognition, and implicit phonological learning on articulation skills of children with Velocardiofacial syndrome (VCFS) and children with cleft palate or velopharyngeal dysfunction (VPD).

Design—Cross-sectional group experimental design.

Participants—8 children with VCFS and 5 children with non-syndromic cleft palate or VPD.

Methods and Measures—All children participated in a phonetic inventory task, speech perception task, implicit priming nonword repetition task, conversational sample, nonverbal intelligence test, and hearing screening. Speech tasks were scored for percentage of phonemes correctly produced. Group differences and relations among measures were examined using nonparametric statistics.

Results—Children in the VCFS group demonstrated significantly poorer articulation skills and lower standard scores of nonverbal intelligence compared to the children with cleft palate or VPD. There were no significant group differences in speech perception skills. For the implicit priming task, both groups of children were more accurate in producing primed nonwords than unprimed nonwords. Nonverbal intelligence and severity of velopharyngeal inadequacy for speech were correlated with articulation skills.

Conclusions—In this study, children with VCFS had poorer articulation skills compared to children with cleft palate or VPD. Articulation difficulties seen in the children with VCFS did not appear to be associated with speech perception skills or the ability to learn new phonological representations. Future research should continue to examine relationships between articulation,

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cognition, and velopharyngeal dysfunction in a larger sample of children with cleft palate and VCFS.

Keywords

Velocardiofacial syndrome; 22q11.2 deletion; articulation; cleft palate; velopharyngeal dysfunction

Velocardiofacial syndrome (VCFS) is a relatively common multianomaly syndrome characterized by cleft palate, velopharyngeal dysfunction, speech-language disorders, cardiac anomalies, cognitive-behavioral disorders, characteristic facial features, differences in brain morphology, and a variety of other health and psychosocial problems (Shprintzen et al., 1978; Shprintzen et al., 1981; Goldberg et al., 1993; Golding-Kushner et al., 1985; Golding-Kushner, 2005; McDonald-McGinn et al., 1997; Eliez et al., 2001). VCFS is a genetic disorder associated with a microdeletion at chromosome 22q11.2, assessed by fluorescent in situ hybridization (FISH) testing (Driscoll et al., 1992; Scambler et al., 1992). Children with VCFS are often initially encountered and diagnosed through a Cleft Palate/Craniofacial Team, likely due to the high incidence of cleft palate, submucous cleft palate, and velopharyngeal dysfunction in this population (Shprintzen et al., 1978; Golding-Kushner et al., 1985; Golding-Kushner, 1991).

Speech disorders in children with VCFS have repeatedly been shown to be more severe and complex than those of children with similar histories of clefting and velopharyngeal dysfunction for speech (VPD) without VCFS (Scherer et al., 1999; Scherer et al., 2001; D'Antonio et al., 2001; DeMarco et al., 2004, 2005). Children with VCFS often display persistent hypernasality and articulation skills that are significantly below those of their age peers. In addition, children with VCFS may demonstrate systematic phonological errors, dysarthria, developmental apraxia of speech, voice disorders, or a combination of these (Kummer et al., 2007; Scherer et al., 1999; Carneol et al., 1999; Persson et al., 2003; Golding-Kushner 1995, 2005). Although children with VCFS may demonstrate speech that is in some ways similar to that of children with non-syndromic cleft palate or VPD, children with VCFS, regardless of the presence of clefting, have consistently been shown to have more impaired articulation skills than these children (Scherer et al., 1999; Scherer et al., 2001; D'Antonio et al., 2001; Golding-Kushner, 1991; DeMarco, 2004, 2005).

While most efforts have focused on describing the speech profile of children with VCFS, few have explored possible causal factors leading to the increased severity of speech disorders in children with VCFS. Researchers have not yet been able to demonstrate a direct link between velopharyngeal dysfunction and articulation skills in the VCFS population, although evidence of some relationship is supported (D'Antonio et al., 2001). In addition, there is very limited research on other factors that might contribute to the increased severity of articulation problems in children with VCFS.

Articulation skills in children with VCFS

Previous research has shown that children with VCFS have reduced consonant inventories and a preference for voiceless consonants regardless of place or manner of articulation, compared to age-matched peers with phenotypic overlap (i.e., similar speech and learning characteristics) but who do not have VCFS or the 22q11.2 deletion (D'Antonio et al., 2001). Persson et al. (2003) and Golding-Kushner (2001, 2005) have reported systematic speech errors similar to the developmental phonological processes often observed in young children with typical speech development. In addition, studies also report a high percentage of glottal stops in the early speech productions of young children with VCFS (D'Antonio et al., 2001;

Scherer et al., 1999; Golding-Kushner, 1991, 2005; Persson et al., 2003). Researchers have not yet found a direct relationship between velopharyngeal factors (e.g., extent of velopharyngeal inadequacy, age at time of palatal repair, type or timing of secondary speech surgery) and measures of articulation skills in children with VCFS (D'Antonio et al., 2001, Scherer et al., 1999). Differences in the ages of participants across studies, as well as variation in speech tasks utilized (e.g. word level vs. spontaneous speech sample) may play a role in explaining at least part of this discrepancy.

The influence of higher-level cognitive-linguistic deficits on articulation skills

Learning disabilities or other language and cognitive impairments are among the most commonly identified characteristics associated with VCFS, with prevalence as high as 99% (Shprintzen et al., 1978; Goldberg et al., 1993; Swillen et al., 1997; Niklasson et al., 2002; Glaser et al., 2002; Goorhuis-Brouwer et al., 2003; Moss et al., 1999; Golding-Kushner et al., 1985; Golding-Kushner, 2005; Campbell & Swillen, 2005). Some previous studies have found a discrepancy between verbal and nonverbal IQ scores in the VCFS population, typically with higher verbal scores (Woodin et al., 2001; Moss et al., 1999; Gerdes et al., 1999; Swillen et al., 1997). Children with significantly decreased cognitive abilities have been found to have more difficulties and delays in processing and producing speech sounds (Eilers and Oller, 1980; Smith and Stoel-Gammon, 1983; Stoel-Gammon, 1997; Iacono, 1998; Kennedy and Flynn, 2002).

Scherer et al. (2001) compared the communication profiles of four children with VCFS to a group of four children with Down syndrome to examine the possible effect of cognitive impairments on speech-language skills. Children with Down syndrome share a common profile with children with VCFS including histories of hypotonia, feeding difficulties, middle ear pathology, and developmental delays (Stoel-Gammon, 1997). Results revealed that the children with VCFS had less developed vocabulary skills and a smaller speech sound inventory compared to the Down syndrome group. So, while children with VCFS may exhibit speech delays or learn aberrant speech production patterns due to velopharyngeal and hearing-related deficits, they may also have more difficulty “unlearning” errors due to specific limitations in cognitive capacity.

The influence of hearing factors and speech perception on articulation skills

Children with VCFS, as well as those with cleft palate or velopharyngeal dysfunction, are at high risk for otitis media with effusion (OME) and subsequent hearing loss (Moller and Starr, 1993; Broen et al., 1996; Digilio et al., 1999; McDonald-McGinn et al., 1997). OME may cause the child to receive an incomplete or distorted auditory signal, due to fluctuating conductive hearing loss. This may then interfere with the development of speech sound perception or discrimination skills (Stool and Randall, 1967; Philips and Harrison, 1969; Heller et al., 1978; Broen et al., 1996) and subsequently, speech production (Finnegan, 1974). Indeed, this relationship is true even in children without craniofacial anomalies: Paden et al. (1987) found that three factors—low articulation scores, elevated hearing thresholds at 500Hz, and a history of early onset and late remission from otitis media with effusion—were the most important variables predicting phonological development in children with otherwise typical development. It is possible that increased persistence and severity of hearing impairment in children with VCFS (Digilio et al., 1999) plays a larger role in the phonological development of children with VCFS than other children with isolated cleft palate and VPD.

The influence of implicit phonological learning ability on articulation skills

Another possible explanation for the increased severity of articulation disorders in children with VCFS is a decreased ability to learn phonological representations for newly

encountered words. Many contemporary theories of speech production (such as the DIVA model, Perkell et al., 2000), posit that auditory-perceptual representations are the targets of speech production. That is, children must accrue information about words' acoustic-perceptual characteristics so that they have well-defined targets in speech production. Hence, a deficit in learning perceptual representations might lead to deficits in expressive phonology. Previous demographic research (i.e., Paden et al., 1987) and experimental research (Munson, Baylis, Krause, & Yim, In Press; Munson, Edwards, & Beckman, 2005) has converged on the notion that an inability to learn phonological representations for novel words underlies the speech production problems of children with phonological disorders (i.e., highly inaccurate speech production in the absence of a clear predisposing condition). It is possible that the articulation errors seen in children with VCFS could be related to a similar perceptual-learning deficit. In addition, if the relationship between phonological learning and speech production learning were seen as reciprocal, then the deficits in articulation in VCFS could also be related to their vocabulary deficits (Scherer et al., 2001).

Phonological learning can be studied experimentally. Fisher et al. (2001) used a long-term auditory priming experiment with preschoolers to investigate this ability. These investigators presented children with spoken nonwords in a *prime* phase, which was followed by a *test* phase in which children repeated nonwords. They found that 2.5 year-old children more accurately identified and repeated nonwords that had been presented twice in the prime phase than nonwords that had not been presented. This result shows that typically developing children can form perceptual representations for words based on minimal exposures. Munson, Baylis, Krause, and Yim (In Press) found that children with phonological disorder show a reduced benefit of priming (brief auditory-perceptual exposure to novel words) in an implicit word-learning task compared to children with typical phonological development. Across the two groups, a statistically significant proportion of variance in the degree of priming (defined operationally as the difference in repetition accuracy between primed and unprimed nonwords) was predicted by a measure of phonetic accuracy in single-word naming, even when chronological age and speech discrimination were controlled statistically. This suggests that speech production abilities are supported by the ability to learn new phonological representations in the implicit priming task.

Overall, there are many complex and interrelated variables which may contribute to the articulation and phonological difficulties in children with VCFS. Studies must take into account factors related to hearing, VPD, cognitive-linguistic abilities, and treatment history. The purpose of this study was to examine the influence of speech sound perception skills, cognitive skills, and implicit phonological learning ability on the articulation skills of children with VCFS and those with cleft palate or VPD. To investigate these factors, we proposed the following hypotheses:

1. Children with VCFS have poorer speech sound perception skills, as measured by accuracy on a minimal pairs discrimination task, compared to children with isolated cleft palate or VPD;
2. Children with VCFS have a disproportionately poorer ability to learn new words, as measured by their articulation skills in an implicit word-learning paradigm, compared to children with isolated cleft palate or VPD; and,
3. Speech perception skills, nonverbal intelligence, and implicit word-learning ability are correlated with articulation skills for both groups of children.

We also attempted to replicate previous research confirming that children with VCFS do have more severe articulation difficulties compared to children with non-syndromic cleft palate, and also expanded our investigation of articulation to include both single word and conversational level phonetic accuracy.

METHOD

Subjects

This study was approved by the University of Minnesota Institutional Review Board and all subjects underwent informed consent procedures and were paid for their participation in the study. Participant demographics are presented in Table 1. Children were recruited through the University of Minnesota Cleft Palate and Craniofacial Clinics by personal or telephone contact, and through a Velocardiofacial Syndrome Family Support Group in the Minneapolis/St. Paul metropolitan area. Parents of all participants completed a Speech Study Parent Questionnaire (Appendix A). This questionnaire was designed for the parent to provide relevant background information on oral-facial structural features, speech therapy history, medical and surgical history, hearing history, and known cognitive-learning factors. The questionnaire was adapted from the University of Minnesota Neuropsychology Clinic Intake Questionnaire to include more speech-specific items and to include elements of the VCFS Specialist Fact Sheet (Velocardiofacial Syndrome Educational Foundation). Selected information from this questionnaire is presented in Table 1.

A total of 13 children (7 females and 6 males) participated in this study and were between the ages of 3;10 to 8;6 (years;months). The VCFS group consisted of five females and three males (ages 4;9–7;0, $M = 6;4$). The comparison group consisted of children with a history of non-syndromic repaired cleft palate, submucous cleft palate, or velopharyngeal dysfunction (referred to as the “CPO/VPD group”). This comparison group consisted of three males and two females (ages 3;10–6;6, $M = 5;6$). The VCFS subjects were, on average, almost one year older than the CPO/VPD group, although this difference did not achieve statistical significance in a Mann-Whitney U Test ($p = 0.171$). All participants were native English speakers in order to rule out confounding effects of second language learning on speech production characteristics and language skills. The CPO/VPD group was selected as an appropriate comparison group for this study because of their phenotypic overlap with children with VCFS. Children in this comparison group would have the following factors in common with the children with VCFS: (1) presence or history of velopharyngeal dysfunction, (2) increased risk of otitis media and fluctuating hearing loss, and (3) increased risk for the development of articulation disorders.

All eight participants in the VCFS group had a 22q11.2 deletion confirmed by fluorescent in situ hybridization testing and clinical diagnosis of VCFS by a geneticist. According to parent report and medical records review, the five children in the CPO/VPD group had (1) no known learning disabilities or mental retardation, (2) no other congenital anomalies or syndromes, (3) no known behavioral or psychiatric conditions, (4) no known language impairments, and (5) no oronasal fistulae at the time of their participation in the study (also confirmed by oral examination). Children with a history of clefting of the primary palate (i.e., involving the lip and alveolus) were excluded from the study to limit interference from dental-occlusal hazards as contributing to articulation errors. Additional information about velopharyngeal and cleft diagnoses and surgical history is presented in Table 1.

Participants were not excluded from the study on the basis of speech therapy or surgical history. While it would have been desirable to control for variables associated with these treatments, it would have been difficult to determine the amount, focus, or therapeutic approach for speech therapy and surgeon or surgery-specific factors for each child. A brief oral examination was also conducted. All speech and hearing testing for this study was conducted in a sound-treated room at the University of Minnesota Department of Speech-Language-Hearing Sciences. All testing was conducted by the first author, a certified and licensed speech-language pathologist.

Global ratings of severity of velopharyngeal inadequacy for speech (VPI) were made from a two-minute audiorecorded portion of each participant's conversational speech sample. Two listeners (the first author and a second rater who works as a speech-language pathologist in a cleft palate clinic) independently rated these samples for type of resonance distortion and overall severity of VPI using an 8-point equal-appearing interval scale. On this scale, 0 indicated no symptoms of VPI and 7 indicated severe symptoms of VPI. Samples were presented in a randomized order. The second listener was not informed of the diagnosis of the participants in the study. The two listeners' severity ratings were averaged and are presented in Table 1. Interlistener reliability for ratings within $\pm$ one scale point was 85%.

All children participated in a brief hearing screening and underwent tympanometry, administered by the first author. Children responded to pure tones at 20dB HL for 500, 1000, 2000, 4000 Hz bilaterally, using a Maico Audiometer (Model MA-20) according to the procedures recommended by the American Speech-Language-Hearing Association (ASHA Panel on Audiologic Assessment, 1996). Tympanometry was also performed according to these ASHA standards. Any participants who did not pass the first hearing screening at 20dB were rescreened again at 25dB. Hearing screening and tympanometry results, as well as information regarding history of otitis media and ventilation tube placement, as obtained from the parent questionnaire, are presented in Table 1. All participants passed the hearing screening at 25dB or better. For the VCFS group, two subjects did not pass the screening at 20dB at two frequencies. However, both of these children did pass a second screening at 25dB. Moreover, both had abnormal tympanometry findings. Neither of these participants had a diagnosed permanent hearing loss, according to parent report. One VCFS participant had negative pressure peaks bilaterally and it was reported in the questionnaire that this was consistent with this participant's history of chronic middle ear dysfunction. The second VCFS participant, who had a reported history of chronic otitis media, had a flattened pressure peak in the right ear, likely due to the presence of fluid in the middle ear space and an extruded ventilation tube present in the ear canal. One child in the CPO/VPD group with no known history of hearing loss also did not pass the hearing screening at 20dB for two frequencies, but did pass at 25dB, and tympanometry revealed patent ventilation tubes. Two children in the CPO/VPD group, both with reported histories of chronic OME, had abnormal tympanometry results indicating slightly negative pressure peaks.

TASKS

Measures of Articulation Skills

Phonetic Inventory (Single-Word Phonetic Accuracy): All children were administered a novel phonetic inventory task in which they named basic items on black-and-white picture cards (see Appendix B for a full list of stimulus words). These single word responses provided a sample of all English phonemes in initial, medial, and/or final word position. The 39 items for this test (including 59 scored consonants and 13 vowels) were selected from existing picture-stimuli based on their phonemic make-up and children's familiarity with the items, as gauged by normative studies by Obler and Albert (1986) and Cyncowicz et al. (1997).

Pictures were presented in semantically uniform sets to facilitate word retrieval and speed of test administration. For example, the stimulus "banana" was followed by "pear," and "pig" was followed by "goat," etc. In the event that a child did not know the name of the pictured item, the examiner had the child produce the correct response after delayed imitation. The responses of this single word articulation inventory were audio-recorded using a Marantz Professional CD Recorder (Model CDR300) and AKG condenser microphone (Model C419IIIIP).

Conversational Speech Sample: Children were given a standard set of toys and parents/caregivers were instructed to play and speak with their child for 15 minutes while a conversational sample was collected. Parents were instructed to limit use of yes/no questions, imitation, and item naming with their child during this time. Speech samples were audiorecorded and later transcribed phonetically by the first author and one independent listener.

The transcribed responses were used to calculate the Percent Phonemes Correct (PPC), using the criteria of Shriberg et al. (1997). The PPC metric expresses the percentage of intended consonant sounds in a speech sample that were articulated correctly. Clinical distortions, deletions, or substitutions of any consonant or vowel are scored as incorrect. Articulation errors were also coded by type and frequency (e.g., substitutions, omissions, distortions, compensatory errors, etc.). For both the single-word picture naming and conversational sample tasks, a separate Percent Phonemes Correct (PPC) score was calculated based on these transcriptions, referred to as PCC-WORD and PCC-CONV, respectively.

Independent Measures

Nonverbal Intelligence: Children were administered the *Kaufman-Brief Intelligence Test* (K-BIT) Matrices and Vocabulary Sections (Kaufman, 1990). The K-BIT is a brief, individually administered measure of verbal and nonverbal intelligence skills. The Vocabulary Subtest measures language development and level of verbal conceptualization as children name black-and-white picture drawings. The Matrices Subtest was included in this study as a nonverbal measure of analogical knowledge using visual stimuli, both meaningful (people and objects) and abstract (designs and symbols). From these test sections, Vocabulary raw scores, Matrices raw scores, and Matrices standard scores and percentile ranks were calculated.

Speech-Sound Perception Skills: To investigate whether children with VCFS have more difficulty detecting the subtle differences between speech sounds, all children participated in a novel minimal-pair identification task. Forty-one sets of minimal pairs of pictures were selected. Stimuli were selected primarily from the same source as the phonetic inventory stimuli, based on their phonetic features and their familiarity to children. These word pairs featured initial and final position phoneme contrasts, selected with the speech characteristics of children with VCFS or cleft palate in mind. For example, the pair “man-pan” was selected because it demonstrates the error of nasal substitution, “boat-goat” was selected for its backing process, and “tie-eye” was selected to resemble a possible glottal stop substitution. Stimuli were produced by an adult male with native Minnesotan dialect (the third author), audio recorded, and later screened for intelligibility before use with the subjects (see Appendix C for a full list of stimulus words).

The child was seated at a table as single auditory stimulus items were presented at 65dB HL using a Dell Latitude D600 laptop, E-Prime software, and two Audix speakers (Model PH5). As each auditory item was presented, a pair of black-and-white picture cards was shown to the child, who was asked to point to the correct response. Responses were scored as correct or incorrect. Overall percentage correct was calculated for each child.

Phonological Learning Ability: The Implicit Priming Task: The implicit priming task explored possible differences in ability to learn new “words” and build phonological representations based on minimal exposure. This was an implicit priming task, modeled on that presented by Fisher et al. (2001). A set of 79 nonwords was created using the phonemes/h, w, p, b, m, n, g, k, t, d, f/, since these are typically the earliest phonemes present in children’s speech repertoire (Smit et al., 1990). These phonemes were combined with vowels to create sequences of CVC and CVCVC nonwords. All of the sequences of

sounds in the nonwords were attested in real English words. For the creation of these stimuli, six adult speakers (3 males, 3 females, all Minnesota natives) were first selected to read aloud this list of nonwords (three times each), and these productions were audio-recorded. These six speakers' productions were then randomized and presented to a set of 5 independent adult listeners with normal speech and hearing using E-Prime software, a laptop computer, and two Audix speakers. Listeners were seated in a soundproof room, presented with the auditory stimulus (the nonword), and then asked to repeat the nonword aloud. The first author then scored the item as intelligible, if the listener produced the word according to the phonetic model, or unintelligible, if the listener's production varied from the phonetically correct model. A percentage of intelligible nonwords was calculated for each of the six speakers and the most intelligible speaker's nonword productions (an adult female) were selected for use in the implicit priming task. Average intelligibility for adults judging the implicit priming stimuli chosen was 99% (see Appendix D for a complete list of implicit priming stimuli).

The procedures for the implicit priming task used with the children were similar to those used by Fisher et al. (2001). In the *prime phase*, children listened to an audio-recording of 52 nonwords which lasted approximately 5 minutes. Each nonword was presented three times in a randomized order for each subject at approximately 65db HL. While listening to the string of nonwords, children were instructed to refrain from talking and color a picture in a coloring book until this recording finished. All participants were compliant with these instructions with the exception of the youngest participant in the CPO/VPD group, who required multiple cues to redirect his attention to the task. After the prime phase, children participated in a two-minute distractor task, which included a brief oral examination and short conversation.

During the *test phase*, children were re-directed to listen to a set of 52 nonwords, one at a time. The test phase took most children, on average, about five minutes to complete. Twenty-six of these nonwords had been presented to the child during the prime phase ("primed nonwords") and twenty-six were novel nonwords ("unprimed nonwords"). After each nonword was presented, the child was asked to repeat each nonword back to the examiner. Children's productions were audio-recorded onto CD-R. Nonword responses were later transcribed to calculate PPC-PRIMED and PPC-UNPRIMED scores for each participant.

A quasi-random representative subset of the Phonetic Inventory and Implicit Priming responses were transcribed and scored by a second independent listener (blind to the diagnosis of the speakers) to assess inter-judge reliability. Reliability for the Phonetic Inventory was calculated at 81.8% overall, with 79.9% agreement for VCFS group responses and 87.0% agreement for the CPO/VPD group's responses. Reliability for the Implicit Priming task was calculated at 83.3%, with 81.5% agreement for VCFS group responses and 90.0% agreement for the CPO/VPD group. Generally, these percentages of agreement are similar to those encountered in other studies utilizing transcription of articulation errors including those associated uniquely with the speech of children with cleft palate (e.g., Estrem & Broen, 1989; D'Antonio et al., 2001; Shriberg et al., 1997). It is likely that there was better reliability for CPO/VPD group due to decreased frequency of errors and decreased severity of articulation impairment. Two Pearson's Chi-Square tests were conducted to determine if these differences in reliability differed significantly between the CPO/VPD and VCFS groups. Neither test was significant ($\chi^2_{[df=1]} = 1.36, p > 0.05$ for the PPC-WORD score; $\chi^2_{[df=1]} = 3.27, p > 0.05$ for the combined Implicit Priming PPC scores).

Data Analysis

Statistical tests used to examine group differences included the Mann-Whitney U-Test and a series of univariate ANOVA's. Spearman's ρ correlation coefficients were calculated to examine relations among measures. An α level of 0.05 was used to determine statistical significance. Scores for the children in each group were summed and averaged for a comparison of group means. In general, nonparametric statistical tests were employed where possible due to the small sample size and exploratory nature of this study.

RESULTS

Articulation Skills

Results for the phonetic inventory and conversational speech sample PPC scores are presented in Table 2. As expected, group differences were statistically significant for the PPC-WORD score, with the children with VCFS demonstrating poorer speech production skills (VCFS: 77.8%, CPO/VPD: 96.4%; $z = -2.13$, $p = 0.03$). Frequency counts and the types of errors produced by the participants are shown in Table 3. Inspection of these data suggest that children with VCFS had a higher frequency of sound substitutions, omissions, compensatory articulation errors, and voicing errors compared to the children with CPO/VPD. Children with VCFS also had lower PPC-CONV scores; however, group differences did not achieve statistical significance at the α level of 0.05 (VCFS: 84.6% CPO/VPD: 93.4% $z = -1.69$, $p = 0.09$). There was no correlation between age and PPC-WORD score (Spearman's $\rho = -0.070$, $p = 0.819$).

Nonverbal Intelligence

For the K-BIT Matrices standard score, group differences in nonverbal intelligence were statistically significant, with the children with VCFS scoring lower than the children with CPO/VPD (VCFS Mean= 88, CPO/VPD Mean=114; $z = -2.49$, $p = 0.011$). Another important finding is that although the children with VCFS, on average, had lower nonverbal intelligence, a mean score of 88 does not classify this group as clinically mentally impaired using conventional criteria, as this score falls within one standard deviation of the normative-sample mean score.

For the K-BIT Vocabulary raw score, group differences were not statistically significant (VCFS: 23, CPO/VPD: 26.8; $z = -.96$, $p = 0.35$) indicating that both groups of children in this study had similar expressive vocabulary skills (see Table 2).

Speech-Sound Perception Skills

Group means for performance on the minimal-pair speech-perception task were very similar (VCFS: 91%, CPO/VPD: 93.8%), suggesting that the groups had comparable speech sound discrimination skills. Rationalized arcsine transformed scores did not significantly differ ($z = -.98$, $p = 0.33$), indicating similar abilities to detect differences among minimal pairs. Qualitative analysis of Tables 1 and 2 suggests the hearing screening status of participants did not appear to affect speech-sound discrimination or articulation performance in this study.

For example, subjects VCFS4, VCFS6, and CPO/VPD4 all passed the screening at 25 dB but outperformed several other subjects who passed at 20 dB (e.g., VCFS1, VCFS5, VCFS8, and CPO/VPD3) on the speech sound perception task. In addition, visual inspection also shows that these subjects who passed at 25 dB had very high PPC-WORD scores (i.e., VCFS4: 95%, VCFS6: 94%, CPO/VPD4: 100%) which were not lower than the rest of the subjects. Unequal group sizes prevented further statistical analysis of these hearing screening data.

Ability to Learn New Words: The Implicit Priming Task

For the PPC-PRIMED score, children with VCFS scored lower than the children with CPO/VPD (VCFS 77.3%, CPO/VPD 90.2%, $z = -1.77$, $p = 0.093$), but this difference did not achieve statistical significance at the α level of 0.05. For the PPC-UNPRIMED score, children with VCFS again had lower scores but the difference was not statistically significant (VCFS 74.5%, CPO/VPD 87.6%; $z = -1.42$, $p = 0.171$). Average PPC scores were higher for primed nonwords than for unprimed nonwords; however, this effect was not statistically significant ($z = -1.508$, $p = 0.132$) for the combined group of 13 participants. Individual participants' performance on the implicit priming task is shown in figure 1. As this figure shows, one child (CPO/VPD5), showed the largest difference between primed and unprimed words in the opposite-than-predicted direction. This child was the youngest to participate in the study and showed high levels of distraction during the prime phase of this task. When results were reanalyzed without this subject's data, a significant effect of priming was detected ($z = -2.207$, $p = 0.027$). This indicates that the primed nonwords were produced more accurately than the unprimed nonwords. The degree of priming was similar for both groups as reflected by a small gain (approximately 3%) in phonetic accuracy PPC scores for the primed nonwords.

Relations among Measures

Nonparametric Spearman's ρ correlation coefficients were utilized in this study to explore relations among measures. The results are presented in Table 4 and show strong correlations ($**p < 0.01$ and $*p < 0.05$) among several measures. In this study, we were most interested in the relationships between measures that might be associated with articulation skills in these groups of children. The correlations between the PPC-CONV and both the PPC-WORD scores ($\rho = 0.920$, $p = 0.01$) and PPC-UNPRIMED scores ($\rho = 0.704$, $p = 0.01$) suggest some relationship between phonetic accuracy at the single word level and the connected speech level. Similar relations were observed between other articulation skill measures, such as between the PPC-WORD scores and both Implicit Priming PPC scores (PPC-PRIMED: $\rho = 0.666$, $p = 0.013$; PPC-UNPRIMED: $\rho = 0.759$, $p = 0.003$). Speech sound perception skills were also related to phonetic accuracy but only for the implicit priming task (PPC-PRIMED: $\rho = 0.655$, $p = 0.015$; PPC-UNPRIMED: $\rho = 0.709$, $p = 0.007$) and not for the single-word phonetic inventory ($\rho = 0.315$, $p = 0.294$).

Age was not correlated with phonetic accuracy (i.e., PPC-WORD, $\rho = -0.070$, $p = 0.819$), likely due to the heterogeneity of articulation skills in the groups of participants. This obviated the need to control for age in our nonparametric correlational analyses. Two factors were found to be significantly correlated with speech production accuracy. First, our measure of nonverbal intelligence (K-BIT Matrices standard score) was highly correlated with phonetic accuracy at the single word and conversational level (PPC-WORD scores: Spearman's $\rho = 0.739$, $p = 0.004$; PPC-CONV scores: $\rho = 0.604$, $p = 0.05$). Nonverbal intelligence was also correlated with performance on the Implicit Priming Task (PPC-PRIMED scores: $\rho = 0.639$, $p = 0.019$; PPC-UNPRIMED scores: $\rho = 0.756$, $p = 0.003$). Second, the severity of VPI was also significantly correlated with speech production accuracy (PPC-WORD scores: $\rho = -0.580$, $p = 0.038$), reflecting that the children with more severe symptoms of VPI were more likely to have poorer articulation skills.

DISCUSSION

Results from this study provide further evidence that the speech phenotype of children with VCFS does demonstrate some overlap with that of children with nonsyndromic cleft palate or velopharyngeal dysfunction; however, we have also identified group differences which may provide insight into the possible nature of each group's speech disorders. As seen in

previous studies (Scherer et al., 1999; D'Antonio et al., 2001), we found that children with VCFS have poorer articulation skills than children with isolated cleft palate or velopharyngeal dysfunction and, on average, more severe ratings of velopharyngeal inadequacy for speech (VPI). Severity of VPI was negatively correlated with the articulation performance of the children in this study, suggesting that the more severe the rating of VPI, the lower the phonetic accuracy score.

In addition to severity of VPI, other treatment factors may have been influencing articulation skills in the VCFS group. Specifically, the two subjects in this study who produced glottal stops (VCFS5 and VCFS7) also had the most severe ratings of VPI. Subject VCFS5 was almost six years of age and had severe articulation difficulties, severe symptoms of VPI, and had not yet received any physical management. In some cases, this could be due to the multiple medical needs that may receive priority over surgical intervention for speech in the more severely affected children with this syndrome. On the other hand, it is also possible that some children with VCFS may have such severe articulation disorders that they interfere with clinicians' ability to diagnose underlying VPI; consequently, appropriate treatment is delayed. Later age of diagnosis and intervention for VPI may account for the more severe articulation difficulties of children with VCFS. Multiple studies of children with cleft palate have found evidence of better velopharyngeal closure and more typical phonological development in children with early palate repair (Dorf and Curtin, 1982; Grobelaar et al., 1995). In addition, it is probable that later diagnosis of VPI is more likely in children with VCFS relative to children with cleft palate, given the large proportion of those children with VCFS without overt clefting. With later treatment, maladaptive articulation behaviors may have already been learned, and these new sound production patterns will have become integrated into the child's developing phonological system. Hence, treatment could be considered "delayed" in many children with VCFS compared to cleft populations. An additional factor worthy of future investigation is the behavioral speech therapy history of children with VCFS, including an examination of duration of therapy, frequency, treatment strategies utilized, and age at which therapy was initiated to determine to what degree earlier and more intensive speech services play a role in the development of articulation skills.

An interesting finding in this study included the observation of higher articulation accuracy at the conversational speech level compared to the single-word level for the VCFS group. This result might suggest that the children with VCFS in this study either (1) employed phonetic avoidances or preferences to maximize intelligibility during conversational speech (e.g., chose to use words with a higher probability of being intelligible to their caregiver), or (2) had restricted expressive vocabularies based on a limited phonetic inventory, thus resulting in obligatory use of a more restricted set of words during the conversational sample. The second hypothesis is not supported by our results, given that we found similar expressive vocabulary levels between the VCFS and CPO/VPD groups in our study. Instead, the phenomenon of phonetic avoidances or preferences has previously been observed in the Down syndrome population (Iacono, 1998). While the phonetic inventory task in our study ensured that all phonemes were produced in multiple word positions, it is possible that in the conversational speech samples, not all phonemes were represented in all positions, resulting in a lack of opportunity for certain target phoneme production. Iacono (1998) hypothesized that children with speech production deficits may demonstrate this discrepancy between conversational and word-level speech samples due to avoidance of certain sounds that they have not mastered, which is also supported by the research of Morrison and Shriberg (1992). It is unclear whether this finding suggests an "active" strategy of phonetic sophistication being utilized by some of the children with VCFS in this study. Given the common finding of relatively stronger verbal than nonverbal intelligence in children with VCFS, this

hypothesis appears plausible and is deserving of further investigation in future research (Woodin et al., 2001; Moss et al., 1999; Gerdes et al., 1999; Swillen et al., 1997).

The children with VCFS in this study did have significantly lower nonverbal intelligence scores than the children with cleft palate or VPD, but their scores were not low enough to be considered clinically mentally impaired. The K-BIT Matrices subtest score depends heavily on visual skills and children with VCFS have previously been shown to have difficulties in this area (Wang et al. 1998; Wang et al., 2000; Simon et al., 2005). Our exploratory analysis of relations among measures revealed a high correlation between nonverbal intelligence and articulation skill measures. There may be a relationship between articulation difficulties in children with VCFS and broader cognitive-linguistic deficits; however, since we did not match our groups for nonverbal intelligence levels, we are unable to determine if this finding is an artifact of having unmatched groups. Future studies should attempt to include a larger group of children with VCFS that represent a wider distribution of cognitive skills, as well as comparison groups matched for cognitive ability, to further examine the influence of nonverbal intelligence on speech performance. This would help provide further insight into whether or how cognitive deficits might be a limiting factor as to why children with VCFS have such significant articulation delays and difficulties.

Regarding the nature of articulation difficulties, this study is the first to provide preliminary information suggesting that children with VCFS may have similar speech sound perception skills compared to children with isolated cleft palate or VPD. The children with VCFS in this study did not demonstrate specific difficulties with a speech sound discrimination task, and performance on this task was not associated with phonetic accuracy. This finding is also important given the history or presence of mild fluctuating hearing loss present in some of the participants. It shows that even in the presence of reduced auditory input, both groups of children were still able to perceive subtle speech sound differences among stimuli.

In addition, no particular deficits were shown by the children with VCFS in the Implicit Priming task. This suggests that there may be no difference between these two groups of children in terms of their ability to learn new phonological representations. Thus, based on the tasks employed in this study, there were no indications to suggest that the articulation difficulties seen in this group of children with VCFS are necessarily related to deficient phonological processing skills or perceptual-learning deficits. Perceptual-learning deficits have recently been suggested to be an endophenotypic marker of phonological impairment in children (Munson et al., in press) and were not found to be characteristic of the group of children with VCFS who participated in this study.

Limitations of this study include the small sample size and an unbalanced group design, which may have reduced the statistical power of our data analysis. This limits our ability to generalize results beyond the groups of children who participated in this study. However, our results lead to some preliminary conclusions and suggest avenues for future research. That is, results from this study suggest that the articulation difficulties seen in children with VCFS may not be related to a specific deficit in speech sound perception or a reduced ability to learn new phonological representations. On the other hand, other types of perceptual-learning processes have not yet been explored in the VCFS population. Neuromotor differences have recently been suggested as a factor influencing articulation difficulties in VCFS (Kummer et al., 2007), but further research is needed to examine how and to what extent VPI affects the articulation profile of children with VCFS. Results of this study lead to additional questions as to how the severity of VPI might interact with the development of articulation skills in VCFS, as well as which speech treatment approaches are most effective for children with this syndrome.

With a better understanding of the underlying factors which contribute to the severe articulation difficulties in VCFS, speech treatment paradigms might be devised to specifically target or compensate for those areas of deficits. Our finding of the lack of group differences in vocabulary, speech sound discrimination, and ability to learn new words supports idea that auditory-verbal aspects of learning may be a relative strength for some children with VCFS, since more consistent deficits have been found in areas of nonverbal and visual-spatial skills. For some children with VCFS, behavioral speech treatment incorporating a combination of traditional visual placement cues and enhanced auditory-perceptual feedback may be effective in facilitating progress with articulation skills. Future research should examine whether there are perceptual-learning strategies and strengths that could be better utilized in behavioral treatment approaches to improve speech outcomes in children with VCFS.

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References

- American Speech Language Hearing Association Panel on Audiologic Assessment. Guidelines for Audiologic Screening. ASHA Desk Reference 1996;IV:333–382.
- Broen PA, Moller KT, Carlstrom J, Doyle S, Devers M, Keenan KM. Comparison of the hearing histories of children with and without cleft palate. *Cleft Palate J* 1996;33:127–136.
- Campbell, L.; Swillen, A. The cognitive spectrum in velo-cardio-facial syndrome. In: Murphy, K.; Scambler, P., editors. *Velo-Cardio-Facial Syndrome: A Model for Understanding Microdeletion Disorders*. Cambridge UP: 2005. p. 147-164.
- Carneol SO, Marks SM, Weik L. The speech-language pathologist: key role in the diagnosis of velocardiofacial syndrome. *Am J Speech Lang Pathol* 1999;8:23–32.
- Cycowicz YM, Friedman D, Rothstein M, Snodgrass JG. Picture naming by young children: norms for name agreement, familiarity, and visual complexity. *J Exper Child Psychol* 1997;65:171–237. [PubMed: 9169209]
- D'Antonio LL, Scherer NJ, Miller LL, Kalbfleisch JH, Bartley JA. Analysis of speech characteristics in children with velocardiofacial syndrome (VCFS) and children with phenotypic overlap without VCFS. *Cleft Palate Craniofac J* 2001;39:455–467.
- DeMarco, AL.; Munson, B.; Moller, KT. Communicative profiles of children with velocardiofacial syndrome and research update. Presented at the annual meeting of the American Speech-Language-Hearing Association; 2004; Chicago, Illinois.
- DeMarco, AL.; Munson, B.; Moller, KT. Predictors of phonetic accuracy in children with 22q11.2 deletion syndrome and children with non-syndromic cleft palate or VPI. Presented at the annual meeting of the American Cleft Palate-Craniofacial Association; 2005; Myrtle Beach, South Carolina.
- Digilio MC, Pacifico C, Tieri L, Marino B, Giannotti A, Dallapiccola B. Audiological findings in patients with microdeletion 22q11.2 (digeorge/velocardiofacial syndrome). *Br J Audiol* 1999;33:329–334. [PubMed: 10890147]
- Dorf DS, Curtin JW. Early cleft palate repair and speech outcome. *Plast Reconstr Surg* 1982;70:74–79. [PubMed: 7089110]

- Driscoll DA, Spinner NB, Budarf ML, McDonald-McGinn DM, Zackai EH, Goldberg RB, Shprintzen RJ, Saal RM, Zonana J, Jones MC, Mascarello JT, Emanuel BS. Deletions and microdeletions of 22q11.2 in velocardiofacial syndrome. *Am J Med Genet* 1992;44:261–268. [PubMed: 1360769]
- Edwards J. Compensatory speech motor abilities in normal and phonologically disordered children. *J Phonetics* 1992;2:189–207.
- Edwards J, Fourakis M, Beckman ME, Fox RA. Characterizing knowledge deficits in phonological disorders. *J Speech Lang Hear Res* 1999;42:169–186. [PubMed: 10025552]
- Edwards J, Fox RA, Rogers CL. Final consonant discrimination in children: effects of phonological disorder, vocabulary size, and articulatory accuracy. *J Speech Lang Hear Res* 2002;45:231–242. [PubMed: 12003507]
- Eilers RE, Oller DK. Comparative study of speech perception in young severely retarded children and normally developing infants. *J Speech Hear Res* 1980;23:419–428. [PubMed: 7442201]
- Eliez S, Blasey C, Schmitt E, White C, Hu D, Reiss A. Velocardiofacial syndrome: are structural changes in the temporal and mesial temporal regions related to schizophrenia? *Am J Psychiatry* 2001;158:447–453. [PubMed: 11229987]
- Estrem T, Broen P. Early speech production of children with cleft palate. *J Speech Hear Res* 1989;32:12–24. [PubMed: 2704186]
- Finnegan DE. Speech sound discrimination skills of seven and eight year old cleft palate males. *Cleft Palate J* 1974;11:111–121. [PubMed: 4524354]
- Fisher C, Hunt C, Chambers K, Church B. Abstraction and specificity in preschoolers' representations of novel spoken words. *J Memory Lang* 2001;45:665–687.
- Gerdes M, Solot C, Wang PP, et al. Cognitive and behavior profile of pre-school children with chromosome 22q11.2 deletion. *Am J Med Genet* 1999;85:127–33. [PubMed: 10406665]
- Glaser B, Mumme DL, Blasey C, Morris M, Dahoun SP, Antonarakis SE, Reiss AL, Eliez S. Language skills in children with velocardiofacial syndrome (deletion 22q11.2). *J Pediatrics* 2002;6:753–58.
- Goldberg R, Motzkin B, Marion R, Scambler PJ, Shprintzen RJ. Velocardiofacial syndrome: a review of 120 patients. *Am J Med Genet* 1993;45:313–319. [PubMed: 8434617]
- Golding-Kushner KJ, Weller G, Shprintzen RJ. Velo-cardio-facial syndrome: language and psychological profiles. *J Craniofac Genet Dev Biol* 1985;5:259–266. [PubMed: 4044789]
- Golding-Kushner, KJ. Unpublished doctoral dissertation. The Graduate School and University Center; New York: 1991. Craniofacial morphology and velopharyngeal function in 4 syndromes of clefting.
- Golding-Kushner, KJ. Treatment of articulation and resonance disorders associated with cleft palate and VPI. In: Shprintzen, RJ.; Bardach, J., editors. *Cleft Palate Speech Management: A Multidisciplinary Approach*. St. Louis: Mosby; 1995. p. 327-351.
- Golding-Kushner, KJ. Speech and language disorders in velo-cardio-facial syndrome. In: Murphy, KC.; Scambler, PJ., editors. *Velo-Cardio-Facial Syndrome: A Model for Understanding Microdeletion Disorders*. Cambridge: Cambridge UP; 2005. p. 181-199.
- Goorhuis-Brouwer SM, Dikkers FG, Robinson PH, Kerstjens-Frederickse WS. Specific language impairment in children with VCFS: 4 case studies. *Cleft Palate J* 2003;40:190–195.
- Grobbeelaar AO, Hudson DA, Fernandes DB, Lentin R. Speech results after repair of the cleft soft palate. *Plast Reconstr Surg* 1995;95:1150–1154. [PubMed: 7761500]
- Heller J, Gens G, Croft C, Moe D. Conductive hearing loss in patients with VPD. *Cleft Palate J* 1978;15:246–253. [PubMed: 278675]
- Iacono T. Analysis of the phonological skills of children with down syndrome from single word and connected speech samples. *Int J Disabil Dev Educ* 1998;45:57–73.
- Kaufman, N. Kaufman-Brief Intelligence Test. Circle Pines, MN: American Guidance Service; 1990.
- Kennedy EJ, Flynn MC. Early phonological awareness and reading skills in children with down syndrome. *Down Syndr: Res Prac* 2002;8:100–109.
- Kummer AW, Lee L, Stutz LS, Maroney A, Brandt JW. The prevalence of apraxia characteristics in patients with velocardiofacial syndrome as compared with other cleft populations. *Cleft Palate Craniofac J* 2007;44:175–181.

- Lamb M, Wilson F, Leeper H. Intellectual function of cleft palate children compared on the basis of cleft type and sex. *Cleft Palate J* 1973;10:367–377. [PubMed: 4517981]
- McDonald-McGinn DM, LaRossa D, Goldmuntz E, Sullivan K, Eicher P, Gerdes M, Moss E, Wang P, Solot C, Schultz P, Lynch D, Bingham P, Keenan G, Weinzimer S, Ming JE, Driscoll D, Clark BJ, Markowitz R, Cohen A, Moshang T, Pasquariello P, Randall P, Emanuel BS, Zackai EH. The 22q11.2 deletion: screening, diagnostic workup, and outcome of results; report on 181 patients. *Genet Testing* 1997;1:99–108.
- Moller, KT.; Starr, CD. *Cleft Palate: Interdisciplinary Issues and Treatment*. Austin: Pro-Ed; 1993.
- Morrison J, Shriberg L. Articulation testing versus conversational speech sampling. *J Speech Hear Res* 1992;35:259–273. [PubMed: 1573866]
- Moss EM, Batshaw ML, Solot CB, et al. Psychoeducational profile of the 22q11.2 microdeletion. *J Pediatr* 1999;134:193–98. [PubMed: 9931529]
- Munson, B.; Baylis, AL.; Krause, MO.; Yim, D. Representation and access in phonological impairment. In: Fougerón, C., editor. *Papers in Laboratory Phonology 10: Variation, Detail, and Representation*. New York: Mouton de Gruyter; In Press
- Munson B, Edwards J, Beckman ME. Relationships between nonword repetition accuracy and other measures of linguistic development in children with phonological disorders. *J Speech Lang Hear Res* 2005;48:61–78. [PubMed: 15938060]
- Unpublished questionnaire. Fairview-University of Minnesota Hospital, Department of Pediatric Neurology; Minneapolis: Neuropsychology Parent Questionnaire.
- Niklasson L, Rasmussen P, Oskarsdottir S, Gillberg C. Chromosome 22q11 deletion syndrome (CATCH-22): neuropsychiatric and neuropsychological aspects. *Dev Med Child Neurol* 2002;44:44–50. [PubMed: 11811651]
- Obler, LK.; Albert, ML. *Action Naming Test*. 1985. Experimental version
- Paden EP, Novak MA, Beiter AL. Predictors of phonologic inadequacy in young children prone to otitis media. *J Speech Hear Dis* 1987;52:232–242.
- Perkell JS, Guenther F, Lane H, Matthies M, Perrier P, Vick J, Wilhelms-Tricarico R, Zandipour M. A theory of speech motor control and supporting data from speakers with normal hearing and with profound hearing loss. *J Phon* 2000;28:233–272.
- Persson C, Lohmander A, Jonsson R, Oskarsdottir S, Soderpalm E. A prospective cross-sectional study of speech in patients with the 22q11.2 deletion syndrome. *J Commun Dis* 2003;36:13–47.
- Philips BJ, Harrison RJ. Language skills of preschool cleft palate children. *Cleft Palate J* 1969;6:108–119. [PubMed: 5253573]
- Richman L. Cognitive patterns and learning disabilities in cleft palate children with verbal deficits. *J Speech Hear Res* 1980;23:447–456. [PubMed: 7442203]
- Scambler PJ, Kelly D, Lindsay E, Williamson R, Goldberg RB, Shprintzen RJ, Wilson DI, Goodship JA, Cross I, Burn J. The velocardiofacial syndrome is associated with chromosome 22 deletions encompassing the digeorge locus. *Lancet* 1992;333:1138–1139. [PubMed: 1349369]
- Scherer NJ, D'Antonio LL, Kalbfleisch JH. Early speech and language development in children with velocardiofacial syndrome. *Am J Med Genet (Neuropsychiatric Genet)* 1999;88:714–723.
- Scherer NJ, D'Antonio LL, Rodgers JR. Profiles of communication disorder in children with velocardiofacial syndrome: comparison to children with down syndrome. *Genet in Med* 2001;3:72–8. [PubMed: 11339384]
- Shprintzen RJ, Goldberg R, Lewin ML, Sidoti EJ, Berkman MD, Argamaso RV, Young D. A new syndrome involving cleft palate, cardiac anomalies, typical facies, and learning disabilities: velocardiofacial syndrome. *Cleft Palate J* 1978;15:56–62. [PubMed: 272242]
- Shprintzen RJ, Goldberg R, Young D, Wolford L. The velocardiofacial syndrome: a clinical and genetic analysis. *Pediatrics* 1981;67:167–172. [PubMed: 7243439]
- Shriberg LD, Austin D, Lewis BA, McSweeney JL, Wilson DL. The percentage consonants correct (PPC) metric: extensions and reliability data. *J Speech Lang Hear Res* 1997;40:708–722. [PubMed: 9263938]
- Smit AB, Hand L, Freilinger JJ, Bernthal JE, Bird A. The Iowa articulation norms and its Nebraska replication. *J Speech Hear Dis* 1990;55:779–798.

- Smith BL, Stoel-Gammon C. A longitudinal study of the development of stop consonant production in normal and down's syndrome children. *J Speech Hear Dis* 1983;48:114–118.
- Simon TJ, Bearden CE, McDonald-McGinn DM, Zackai E. Visuospatial and numerical cognitive deficits in children with chromosome 22q11.2 deletion syndrome. *Cortex* 2005;41:131–141.
- Stoel-Gammon C. Phonological development in down syndrome. *Mental Retardation Dev Disabil Res Reviews* 1997;3:300–306.
- Stool SE, Randall P. Unexpected ear disease in infants with cleft palate. *Cleft Palate J* 1967;4:99. [PubMed: 5229150]
- Swillen A, Devriendt K, Legius E, Eyskens B, Dumoulin M, Gewillig M, Fryns JP. Intelligence and psychosocial adjustment in velocardiofacial syndrome: a study of 37 children and adolescents with VCFS. *J Med Genet* 1997;34:453–458. [PubMed: 9192263]
- Velocardiofacial Syndrome Specialist Fact Sheet. Velocardiofacial Syndrome Educational Foundation; <http://www.vcfsef.org/AboutVCFS/FactSheet/factsheet.pdf>
- Wang PP, Solot C, Moss EM, Gerdes M, McDonald-McGinn DM, Driscoll DA, Emanuel BS, Zackai EH. Developmental presentation of 22q11.2 deletion (diGeorge/velocardiofacial syndrome). *J Dev Behav Pediatrics* 1998;19:342–345.
- Wang PP, Woodin MF, Kreps-Falk R, Moss EM. Research on behavioral phenotypes: velocardiofacial syndrome (deletion 22q11.2). *Dev Med Child Neurol* 2000;42:422–427. [PubMed: 10875531]
- Woodin M, Wang PP, Aleman D, McDonald-McGinn DM, Zackai E, Moss E. Neuropsychological profile of children and adolescents with the 22q11.2 microdeletion. *Genet Med* 2001;3:34–39. [PubMed: 11339375]

Appendix A. Parent Questionnaire

VELOCARDIOFACIAL SYNDROME RESEARCH STUDY QUESTIONNAIRE*

Date of birth.: _____ Gender: Male Female

Today's date: _____

Person filling out this form: Mother Father Other: _____

Primary language spoken by this child: _____

Which of the following conditions, if any, has your child been formally diagnosed with?

_____ Velocardiofacial syndrome

_____ DiGeorge syndrome

_____ none of these

Result of FISH test: _____ positive for 22q11.2 deletion (confirmed by medical records)

_____ negative (no deletion)

Facial/Oral Findings

Has this child been diagnosed as having any of the following conditions?

_____ Cleft palate

_____ Submucous cleft palate (age at diagnosis: _____)

_____ Cleft lip only

_____ Cleft lip and cleft palate

_____ No cleft diagnosed but “velopharyngeal dysfunction,” “velopharyngeal inadequacy for speech,” “VPI,” or “hypernasal speech”

At what age was this diagnosed? _____

Ear/Hearing Findings

Has this individual had ear or hearing-related problems? Yes/No

Which type? (please check all that apply)

_____ Frequent ear infections or fluid in ears

Did this child require placement of tubes in their ears? Yes/No

At what age were tubes placed? _____

_____ Hearing loss (Please specify: _____)

_____ Other: _____

Surgical History

Please check which of following procedures this child has had performed:

_____ palate repair: age _____

_____ pharyngeal flap: age _____

_____ sphincter pharyngoplasty: age _____

_____ tonsils removed: age _____

_____ adenoids removed: age _____

_____ heart surgery: age _____

_____ Other hospitalizations, illnesses, and surgeries: _____

Speech/Language History

Has your child been diagnosed with a communication disorder? Yes/No

Age what age was this problem identified? _____

Please check which of the following your child has been diagnosed with:

_____ delay in speech sound development

_____ delay in expressive language development

_____ delay in receptive language development

_____ articulation disorder

_____ phonological disorder

- _____ apraxia of speech (difficulty with coordination or sequencing of speech sounds)
- _____ language disorder (difficulty with forming sentences, concepts, grammar, syntax, word finding)
- _____ voice disorder (hoarseness, breathy, strained voice, vocal nodules, etc.)
- _____ hypernasal speech (too much nasality in speech, associated with VPI)
- _____ stuttering
- _____ social interaction difficulties
- _____ Other: _____

Has this child received speech therapy? Yes/No

At what age did they start speech therapy? _____

How many years have they received speech services? _____

Cognitive/Learning/Behavioral

Has this individual received a formal diagnosis of any of the following:

_____ Learning disabilities

In which areas (e.g., math, reading, spelling, etc.)? _____

_____ Cognitive impairment or mental retardation

_____ ADHD (Attention deficit hyperactivity disorder) or ADD

_____ Nonverbal learning disability

_____ Auditory Processing disorder (APD or CAPD)

_____ Autistic Spectrum disorder:

_____ Asperger's _____ PDD (pervasive dev. disorder) _____ Autism

_____ Obsessive-compulsive disorder

_____ Depression

_____ Anxiety Disorder

*This is an abbreviated version of the original questionnaire provided to parents. This version includes those questions most relevant to the data reported in this study, seen in Table 1.

Figure 1. Percent phonemes correctly repeated for unprimed and primed words in the implicit priming task, separated by individual subject.

Table 1

Information for the 13 participants obtained from the Parent Questionnaire, medical records review, and hearing screening.

Subject	Sex	Age	VP Dx	VP Surgical Hx	Resonance	VPI Severity	Speech Tx (yrs)	OME Hx	Hearing	Tubes	Tymps
VCFS1	F	4; 11	SMCP	none	hyper	1.5	4	no	dB 20	No	WNL
VCFS2	M	7; 6	VPD	none	hyper	5.5	5	yes	dB 20	No	NPP
VCFS3	F	6; 11	VPD	PH FLAP	hyper	2	6	yes	dB 20	No	WNL
VCFS4	F	7; 6	SMCP	PH FLAP	hyper	3	7	no	dB 25	No	NPP
VCFS5	M	5; 11	VPD	none	hyper	7	3	no	dB 20	No	WNL
VCFS6	F	6; 2	SMCP	PH FLAP	hypo	0	2	yes	dB 25	Yes	FPP
VCFS7	F	7; 5	SMCP	SPHINCTER	hyper	6	7	yes	dB 20	No	WNL
VCFS8	M	4; 9	CP	SPHINCTER	hyper	3.5	3	yes	dB 20	No	WNL
CPO/VPD1	F	5; 9	SMCP	none	hyper	1	3	no	dB 20	No	NPP
CPO/VPD2	M	6; 5	CP	none	hypo	0	1	yes	dB 20	No	NPP
CPO/VPD3	M	6; 6	VPD	none	hyper	2	3	yes	dB 20	No	WNL
CPO/VPD4	F	4; 8	CP	none	hyper	1.5	0	yes	dB 25	Yes	WNL
CPO/VPD5	M	3; 10	VPD	none	hyper	1	1	yes	dB 20	Yes	WNL

Note **AGE** is presented in years:months. **VP Dx** = Initial velopharyngeal diagnosis: **CP** = cleft palate, **SMCP** = submucous cleft palate, **VPD** = other velopharyngeal dysfunction (non-cleft, e.g., post-adenoidectomy). **VP Surgical Hx** = velopharyngeal speech surgery history: **PH FLAP** = pharyngeal flap, **SPHINCTER** = sphincter pharyngoplasty. **Resonance** = Current type of resonance distortion, **hyper** = hypernasality, **hypo** = hyponasality. **VPI Severity** = global rating of severity of velopharyngeal inadequacy for speech (based on conversational sample using 8 point EAI scale rating, 0 = no current symptoms of VPI, 7 = severe velopharyngeal inadequacy for speech). **Speech Tx** = Years of speech therapy. **OME Hx** = history of chronic OME. **Hearing** = hearing screening pass level. **Tubes** = current presence of ventilation tubes. **Tymps** = tympanometry results: **WNL** = within normal limits, **NPP** = negative pressure peaks, **FPP** = flattened pressure peaks.

Table 2

Participant scores for all tasks

Subject	AGE (yrs; mos)	K-BIT V-RAW	K-BIT M-RAW	K-BIT MSS	SPEECH PERCEP	PPC-WORD	PPC-PRIMED	PPC-UNPRIMED	PPC-CONV
VCFS1	4;11	11	7	73	84	83	66	58	86
VCFS2	7;6	32	16	84	98	92	92	84	94
VCFS3	6;11	26	16	92	95	90	88	87	94
VCFS4	7;6	26	20	97	95	99	92	87	95
VCFS5	5;11	16	14	92	88	25	39	34	49
VCFS6	6;2	26	18	102	100	94	85	91	94
VCFS7	7;5	32	19	96	98	64	88	82	78
VCFS8	4;9	15	6	68	70	75	68	73	87
Mean		23	15	88	91	78	77	75	85
S.D.		8	5	12	10	24	19	19	16
CPO/VPD1	5;9	23	14	94	98	92	91	84	88
CPO/VPD2	6;5	36	33	156	98	97	98	90	95
CPO/VPD3	6;6	33	23	112	100	96	96	90	89
CPO/VPD4	4;8	27	14	105	98	100	92	90	99
CPO/VPD5	3;10	15	12	103	75	97	74	84	96
Mean		27	19	114	94	96	90	88	93
S.D.		8	9	24	11	3	9	3	5

K-BIT VRAW = K-BIT Vocabulary section raw score; **K-BIT MRAW** = K-BIT Matrices section raw score; **K-BIT MSS** = K-BIT Matrices standard score (mean = 100, 50th percentile); **SPEECH PERCEP** = percentage correct on minimal pairs perception task; **PPC-WORD** = Single-word picture naming phonetic accuracy (percent phonemes correct); **PPC-PRIMED** = Implicit Priming Task primed words phonetic accuracy (percent phonemes correct); **PPC-UNPRIMED** = Implicit Priming Task unprimed words phonetic accuracy (percent phonemes correct); **PPC-CONV** = Conversational speech sample phonetic accuracy (percent phonemes correct).

Table 3

Frequency and types of articulation errors from the phonetic inventory task.

SUBJECT	SUBSTIT	OMISS	GLOTTAL	NFRIC	DISTORT	VOICING	WEAK PR	ANE
VCF51	9	3	0	0	1	3	2	0
VCF52	1	5	0	0	1	1	9	0
VCF53	6	1	0	0	0	3	0	1
VCF54	1	0	0	0	0	0	3	0
VCF55	34	11	16	6	0	0	2	5
VCF56	3	1	0	0	4	2	0	0
VCF57	25	1	2	0	1	0	9	3
VCF58	15	0	0	0	0	0	0	0
CPO/VPD1	5	1	0	2	6	0	2	2
CPO/VPD2	2	0	0	0	0	0	0	0
CPO/VPD3	2	1	0	0	2	0	2	10
CPO/VPD4	0	0	0	0	0	0	0	0
CPO/VPD5	2	1	0	0	0	0	0	0

Frequency counts above reflect raw number of errors for each error type based on words produced during phonetic inventory task. SUBSTIT = Total # of substitutions (including compensatory errors), OMISS = # omissions, GLOTTAL = # of glottal stop substitutions, NFRIC = # of nasal fricative substitutions, DISTORT = # of distortions (placement-related distortions only), VOICING = # of voicing errors (i.e., devoicing), WEAK PR = # of weak pressure consonants, ANE = # of occurrences of audible nasal air emission (does not include nasal fricatives).

Table 4Correlations (Spearman's ρ) among Measures

	PPC-CONV	K-BIT VRAW	K-BIT MSS	SPEECH PERCEP	PPC-WORD	PPC-PRIMED	PPC-UNPRIMED
PPC-CONV	1.000						
K-BIT VRAW	0.285	1.000					
K-BIT MSS	0.604*	0.599*	1.000				
SPEECH PERCEP	0.156	0.777**	0.563*	1.000			
PPC-WORD	0.920**	0.350	0.739**	0.315	1.000		
PPC-PRIMED	0.549	0.855**	0.639*	0.655*	0.666*	1.000	
PPC-UNPRIMED	0.704**	0.620*	0.756**	0.709**	0.759**	0.702**	1.000

K-BIT VRAW = K-BIT Vocabulary section raw score; K-BIT MRAW = K-BIT Matrices section raw score; K-BIT MSS = K-BIT Matrices standard score; SPEECH PERCEP = percentage correct on minimal pairs speech sound discrimination task; PPC-WORD = Single-word phonetic inventory percent phonemes correct; PPC-PRIMED = Implicit Priming primed words percent phonemes correct; PPC-UNPRIMED = Implicit Priming unprimed words percent phonemes correct; PPC-CONV = Conversational speech sample percent phonemes correct.

* $p < .05$ and

** $p < .01$.

Appendix B

Stimuli for the phonetic inventory, with broad phonetic transcriptions.

Word	Transcription	Word	Transcription
<i>banana</i>	[bəˈnænə]	<i>thumb</i>	[θʌm]
<i>lemon</i>	[ˈlemən]	<i>teeth</i>	[tiθ]
<i>carrot</i>	[ˈkæ.ɹɪt]	<i>whistle</i>	[ˈwɪsəl]
<i>potato</i>	[pəˈteɪtoʊ]	<i>watch</i>	[wɒtʃ]
<i>cookie</i>	[ˈkʊki]	<i>glove</i>	[glʌv]
<i>gum</i>	[gʌm]	<i>zipper</i>	[ˈzɪpə]
<i>cat</i>	[kæt]	<i>matches</i>	[ˈmætʃɪz]
<i>elephant</i>	[ˈɛlɪfənt]	<i>scissors</i>	[ˈsɪzəz]
<i>giraffe</i>	[dʒəˈæf]	<i>shovel</i>	[ˈʃʌvəl]
<i>pig</i>	[pɪg]	<i>ladder</i>	[ˈlædə]
<i>sheep</i>	[ʃip]	<i>table</i>	[ˈteɪbəl]
<i>chicken</i>	[ˈtʃɪkən]	<i>vase</i>	[veɪs]
<i>duck</i>	[dʌk]	<i>house</i>	[haʊs]
<i>feather</i>	[feð]	<i>bridge</i>	[brɪdʒ]
<i>cages</i>	[ˈkeɪdʒɪz]	<i>wagon</i>	[ˈwæɡən]
<i>fish</i>	[fɪʃ]	<i>moon</i>	[mun]
<i>fishing</i>	[ˈfɪʃɪŋ]	<i>web</i>	[web]
<i>boy</i>	[bɔɪ]	<i>red</i>	[ɹed]
<i>nose</i>	[noʊz]	<i>those</i>	[ðoʊz]

Appendix C

Stimulus pairs for the minimal-pairs speech perception task.

Pair	Contrast	Pair	Contrast
pan	/p-/m/	kite	/k-/n/
pear	/p-/h/	goat	/g-/k/
panis	/p-/Ø	mail	/m-/w/
purse	/p-/n/	nose	/n-/h/
peel	/p-/w/	knot	/n-/h/
bat	/b-/h/	farm	/f/Ø
bee	/b-/k/	fan	/f-/k/
bone	/b-/f/	fire	/f-/t/
book	/b-/k/	fox	/f-/s/
bear	/b-/p/	sail	/s-/n/
boat	/b-/g/	sew	/s-/t/
bite	/b-/k/	sand	/s-/h/
tie	/t-/Ø	shoe	/ʃ-/z/
tail	/t-/n/	wig	/w-/p/
top	/t-/m/	spit	/sp-/s/
dog	/d-/h/	ski	/sk-/s/
deer	/d-/Ø	swing	/sw-/s/
cat	/k-/h/	snail	/sn-/s/
cap	/k-/m/	scale	/sk-/m/
comb	/k-/h/	stool	/st-/t/
cook	/k-/h/	chair	/tʃ-/h/
jacks	/dʒ-/Ø	cup	/p-/t/
bat	/t-/g/	rope	/p-/d/
lock	/k-/g/	mouth	/θ-/s/
leaf	/f-/k/	wash	/ʃ-/tʃ/
gun	/n-/m/	soap	/p-/w/
pipe	/p-/Ø	ice	/s-/Ø
beach	/tʃ-/Ø	time	/m-/Ø

“Ø” indicates the omission of a consonant.

Appendix D

Stimuli for the implicit priming experiment, transcribed phonetically.

PRIMED STIMULI		UNPRIMED STIMULI	
[bug]	[kʌtəm]	[bæn]	[kænid]
[bukɛf]	[kɪ dəm]	[bøkæm]	[kɛnid]
[fæfeɪb]	[nædəm]	[dɪt]	[mudub]
[fub]	[nek]	[dɪnəp]	[næf]
[gæɡʊt]	[pɛd]	[fudup]	[pæb]
[ɡɪb]	[pɛnɪn]	[ɡɪf]	[pætɪk]
[hʌ g æb]	[tɛn]	[ɡɪɡɛɪm]	[pɛk]
[jæbeɪ g]	[tɔɡ]	[hɪmɔt]	[tɪmæk]
[jæg]	[tʌmud]	[jæf]	[tɪp]
[jeɪf]	[wɛdɪt]	[jɛb]	[wæfug]
[jun]	[wɛk]	[jʌɡɪf]	[wʌb]
[judɪk]	[wuk]	[jum]	[wɔdɪk]
[kʌn]	[wupɛd]	[kæk]	[wɔk]