Analysis of Speech Characteristics in Children With Velocardiofacial Syndrome (VCFS) and Children With Phenotypic Overlap Without VCFS

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Objective: **To address two questions of theoretical importance regarding the profile and course of communication impairment associated with velocardiofacial syndrome (VCFS): (1) do speech characteristics of children with VCFS differ from a group of children with some of the phenotypic characteristics of VCFS who do not have the syndrome, and (2) do younger children with VCFS demonstrate speech patterns that differ from older children with VCFS?**

Design: **Prospective, cross-sectional study comparing two groups of children at two age levels.**

Patients: **Thirteen children with VCFS and eight children with some of the phenotypic features of VCFS who did not have the syndrome. Children ranged in age from 3 to 10 years.**

Main Outcome Measure: **(1) Broad phonetic transcription of speech yielding measures of number of consonant types, Percent Consonant Correct, and percentage of glottal stops used; and (2) composite ratings of velopharyngeal function made from perceptual, aerodynamic, and endoscopic evaluations.**

Results: **Younger children with VCFS demonstrated greater speech impairment than older children with VCFS or the children without VCFS, such as smaller consonant inventories, greater number of developmental errors, greater severity of articulation disorder, and higher frequency of glottal stop use. The relationship between ratings of velopharyngeal function and the speech variables analyzed was not straightforward.**

Conclusions: **Some young children with VCFS demonstrated speech impairment that is qualitatively and quantitatively different from older children with VCFS or children without VCFS. This finding supports the hypothesis that some children with VCFS demonstrate a profile of speech production that is different from normal but also may be specific to the syndrome.**

KEY WORDS: *cleft palate, speech, 22q11 deletion, velocardiofacial syndrome*

Velocardiofacial syndrome (VCFS) is a common multianomaly syndrome first described by Shprintzen and colleagues in 1978 (Shprintzen et al., 1978). More than 180 clinical features have been described with the most common features being; characteristic facies, conotruncal heart anomalies, palatal clefting (overt, submucous, or occult submucous), learning disabilities, and behavioral disorders (Shprintzen et al., 1981; Goldberg et al., 1993; McDonald-McGinn et al., 1997; Shprintzen, 1998). The syndrome has been associated with a submicroscopic deletion at chromosome 22q11.2 that can be confirmed by fluorescent in situ hybridization (FISH; Scambler et al., 1992; Driscoll et al., 1992a, 1992b; Morrow et al., 1995).

Speech and language impairment is a prominent feature of the syndrome. Shprintzen et al. (1978) first described the speech and resonance characteristics of children with VCFS. Golding-Kushner et al. (1985) presented a description of language patterns in a group of 26 children with VCFS identifying significant language impairment in their population of children. Gerdes et al. (1999) reported several measures of cognitive function and behavior in a large population of pre-

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school children with VCFS. Reports of speech and language measures were available for a subset of the greater population showing a wide range of speech and language abilities from normal to significantly delayed. The authors reported that 62.5% of the children tested were generally nonoral communicators at 24 months of age, and these delays in language were beyond what would be expected for their developmental level. However, the speech and language data were a small portion of the overall report, and data regarding the profiles of communication impairment or individual data were not presented. Similarly, Moss et al. (1999) reported speech and language data pertaining to a group of school-age children and showed standard language scores were below verbal IQ and were commensurate with performance IQ. These data suggest that the profile of communication impairment in the school-age population described by Moss et al. differs from the profile found for the preschool children in the population described by Gerdes et al. (1999). It is unclear whether these differences represent a developmental trend of differences in the populations or the testing methods.

All of these studies utilized group data for standardized tests that are commonly used in clinical practice. However, few research data are available in the literature that utilizes accepted methodologies for analyzing speech-language test data in such a way that allows for comparison with published studies of speech and language development in other clinical populations. Also, all of the studies of speech and language performance in children with VCFS have utilized different test measures making comparison among studies difficult. Therefore, one area that has been lacking in the reports of communication deficits in children with VCFS is a methodology for consistency of analysis that is accepted in the speech and language development literature and therefore allows for comparison across study groups and different ages.

Scherer et al. (1999, 2001) reported two studies of communication development in children with VCFS that utilized standard metrics following a developmental approach. For example, in the existing literature, speech patterns in children with VCFS have been described using simple scores on articulation tests or qualitative descriptions of compensatory errors. Scherer et al. (1999, 2001) demonstrated the benefit of narrow phonetic transcription, whole word transcription, and error analysis with normative comparisons when attempting to understand some of the patterns of speech production frequently observed in children with VCFS. Although speech, language, and learning disorders are common in the syndrome, there has been little theoretical discussion or hypothesis development in the literature regarding the origin and course of the communication deficits associated with VCFS. Furthermore, little information has been published regarding the most appropriate treatment strategies for communication disorders associated with VCFS (Golding-Kushner, 1995; Kok and Solman, 1995; Golding-Kushner and Shprintzen, 1998; Scherer and D'Antonio, 1998; Solot et al., 1998).

Scherer et al. (1999) reported an in-depth, longitudinal study describing speech and language development in a sample of four children with VCFS from 6 months to 30 months of age. Performance of the children with VCFS was contrasted with the performance of three comparison groups: normally developing children, children with cleft lip and palate, and children with isolated cleft palate. Young children with VCFS showed receptive-expressive language impairment from the onset of language. Speech and expressive language development for the children with VCFS were severely delayed beyond a level predicted by general development or receptive language performance. The children with VCFS showed severe limitations in speech sound inventories and deficits in early vocabulary development that exceeded those delays demonstrated by children with cleft lip and palate or children with isolated cleft palate. For the children with VCFS, early vocabulary and speech sound acquisition were severely impaired, leaving them essentially nonoral communicators through 30 months of age.

An important feature of the study by Scherer et al. (1999) was the data concerning the relationship between speech production and velopharyngeal (VP) function. This was the first comparison of speech production skills of children with VCFS to groups of children with palatal clefting. Many of the children with VCFS have overt cleft palate, submucous cleft, or occult submucous cleft, and it has been assumed that velopharyngeal inadequacy (VPI) is a significant contributor to the speech production patterns observed in these children. Thirty percent to 84% of individuals with VCFS have been reported to demonstrate symptoms of VPI (McDonald-McGinn et al., 1997; Nayak and Sell, 1998). Also, the speech patterns of children with VCFS have been described as having a predominance of glottal stop substitutions for whole classes of sounds. Glottal stops frequently occur in children with cleft palate as a compensation for VPI. It has been suggested that the high occurrence of glottal stop substitutions is responsible for much of the oral communication impairment in young children with VCFS. Data from the study by Scherer et al. (1999) did not support a simple relationship between the severe speech production abnormalities observed for the young children with VCFS and the presence of VPI. The VCFS group demonstrated significantly greater speech production deficits than children in the two cleft groups who also experienced VPI. The authors concluded that in the young children studied, the relationship between VP function and speech sound errors was not as simple and straightforward as has been suggested previously.

Scherer et al. (1999) described several areas of practical and theoretical importance regarding the speech and language impairments associated with VCFS that warranted further investigation. Results from their study point to a distinctive profile of communication impairment for young children with VCFS that differs in severity and pattern from commonly identified profiles of speech and language impairment. The authors raised several questions that remain unanswered. Are there subgroups within the profile of speech and language impairment described for children with VCFS? Do profiles of communication impairment change with increasing age after 30 months or with treatment? What is the relationship between VPI and the type and severity of speech sound errors?

Purpose

To address some of the questions that exist regarding the speech disorders associated with VCFS, there are a variety of methodologic issues that should be considered. First, methodologies should be employed that utilize standard research metrics for descripting speech development in a manner that allows for comparison to the normal speech development literature and the research literature regarding other clinic populations. Another important methodologic issue pertains to identification of appropriate comparison groups. Depending on the question being asked, different comparison groups are needed. For example, in a study of the frequency of speech and language impairment, comparison groups might include craniofacial populations and cardiac populations as well as general pediatric populations. To address the question regarding the effects of clefting on speech and language development in children with VCFS, the use of cleft and noncleft comparison groups is appropriate. One important question addresses whether the profile of speech and language impairment demonstrated by children with VCFS is unique. Preliminary data concerning this question would be obtained by comparing speech and language profiles of children with VCFS with a group of children with speech and language deficits who have some of the phenotypic characteristics of VCFS but who do not have the syndrome.

Another issue that raises important questions regarding methodology is the study of the course of speech and language impairment over time. The cross-sectional and case studies in the literature have not addressed the developmental course of the communication symptoms. The most important variables that should be considered in longitudinal studies have not been delineated. Studies of cross-sectional populations in which the data are analyzed for temporal trends are needed. Results from such studies can be utilized to provide preliminary data identifying critical variables for future longitudinal studies.

Two specific questions were addressed in the present study. First, are the speech production characteristics of children with VCFS different from a group of children with speech impairment who present with some of the phenotypic characteristics of VCFS but who do not have the syndrome? These data will provide information pertaining to the broader question regarding whether the profile of speech characteristics displayed by children with VCFS is specific to the syndrome. The second specific question is, Do younger children with VCFS demonstrate the same speech patterns as older children with VCFS? Data from this cross-sectional analysis of different age groups may assist in the generation of hypotheses regarding the developmental course of speech patterns in children with VCFS.

Method

Subjects

Twenty-one children served as subjects for this study. All children were referred for comprehensive evaluation of speech production and VP function following screening by a regional Cleft Palate/Craniofacial Team. Each child demonstrated abnormal speech production patterns and some speech characteristics that were suggestive of VPI. Each had several features consistent with a diagnosis of VCFS. FISH analysis using DNA probes for 22q11.2 was done on a lymphocyte karyotype of each child.

Because some children with VCFS may not show a deletion that can be identified with routine FISH analysis, the diagnosis was further confirmed by an independent examiner. Videotaped images of the children were provided to an expert in the diagnosis of VCFS. Results of the FISH were unknown to the expert who was asked to identify which children had phenotypic craniofacial features consistent with a diagnosis of VCFS. Following the results of the FISH and the assessment of phenotypic characteristics, the children were divided into two groups. The first group was comprised of the children with phenotypic characteristics of VCFS and a deletion at 22q11 and are referred to as the "VCFS group." The second group was comprised of the children who had some of the phenotypic characteristics of VCFS but who did not have a deletion at 22q11 and who were judged by an expert not to have the syndrome. For purposes of this study, the latter group is referred to as the "comparison group."

Subjects ranged in age from 3 years 4 months to 10 years 2 months. There were 13 children (11 girls and 2 boys) in the VCFS group and 8 children (6 girls and 2 boys) in the comparison group. A chart review documented the subjects' medical and developmental histories. The history form utilized was the Velocardiofacial Specialist Fact Sheet published by the VCFS Educational Foundation (Shprintzen, 1998). Table 1 shows the age, sex, and cleft type of each subject. Table 2 shows the physical and developmental features of the VCFS group and the comparison group.

In the normal progression of speech development, speech sound accuracy generally approaches the adult model by the age of 7 years. Therefore, many studies of speech disorders in children divide subjects above and below this developmental boundary (Shriberg et al., 1997a). In addition, there are anecdotal reports and suggestions among clinicians experienced in treating children with VCFS that there are differences in the type and severity of communication deficits demonstrated by younger and older children with VCFS (Shprintzen, personal communication). Therefore, for purposes of this study, data analysis was performed for two age groups. The younger group was comprised of children 6 years 11 months and under, and children over 7 years comprised the older group.

Procedures

Phonetic Transcriptions

Speech productions were examined through phonetic transcriptions of videotaped speech stimuli. The videotaped speech sample was collected during the clinical evaluation of speech and VP function and included a brief conversational speech

TABLE 1 Chronological Ages for Children in the Velocardiofacial Syndrome (VCFS) and Comparison Groups*

	VCFS				*Comparison*			
	Subject No.	*Age, y, mo*	*Cleft Type*	*Sex*	*Subject No.*	*Age, y, mo*	*Cleft Type*	*Sex*
Younger	1	3, 4	CP	F	1	5, 2	NC	F
	2	4, 4	OSMCP	F	2	6, 4	NC	F
	3	4, 8	SMCP	M	3	6, 7	CP	F
	4	5, 6	UCLP	F	4	6, 7	VPDYS	M
	5	5, 8	OSMCP	M	Mean	6, 2		
	6	5, 11	NC	F				
	7	6, 1	SMCP	F				
	Mean	5, 1						
Older	8	7, 3	SMCP	F	5	7, 7	SMCP	M
	9	7, 7	SMCP	F	6	8, 1	OSMCP	F
	10	7, 6	VPDYS	F	7	8, 6	SMCP	F
	11	9, 9	OSMCP	F	8	9, 6	SMCP	F
	12	10, 2	SMCP	F	Mean	8, 5		
	13	11, 0	OSMCP	F				
	Mean	8, 11						

* CP = cleft of secondary palate; OSMCP = occult submucus cleft palate, SMCP = submucous cleft palate; UCLP = unilateral cleft lip and palate, VPDYS = velopharyngeal dysproportion; NC = noncleft, F = female; M = male.

sample and repetitions of modeled words and sentences. The speech stimuli included 18 single words containing all consonant place and manner of articulation features in initial and final position with varying vowel contexts. The connected speech samples included sentences that were controlled for place and manner of articulation features and spontaneous conversation. A broad phonetic transcription using diacritics for nasality and compensatory articulation patterns was completed by a single transcriber trained extensively in use of the International Phonetic Alphabet for transcribing the speech of children with disordered articulation and VP symptoms. Particular emphasis was placed on the accurate transcription of compensatory articulation errors, particularly glottal stops as described previously (Scherer et al., 1999).

Reliability

Phonetic transcriptions of disordered speech are subject to methodologic considerations that can reduce the validity of the method. However, Shriberg et al. (1997b) have described procedures for reducing these methodologic limitations, and these suggestions were incorporated in the present study. Specifically, transcribers received training sessions reviewing transcription notations and periodic transcription practice. Transcribers maintained 85% reliability on practice transcriptions.

Intrarater reliability was calculated for the transcriber using 20% of the original speech sample approximately 1 month following the initial transcription. Point-by-point intrarater reliability was 92% agreement for the repeated transcriptions. Interrater reliability was computed for 100% of the single words and short sentences from the videotaped evaluations transcribed by a second transcriber with training similar to the primary transcriber. An average percent agreement was calculated for consonant productions. Point-by-point interrater reliability was calculated to be 82% agreement, which compares favorably with reliability measures reported previously for transcriptions of disordered speech (Shriberg et al., 1997b). In the cases of disagreement, a third transcriber was used to break the disagreements between the two transcribers. If the third transcription agreed with one of the two original transcribers, then that transcription was used in the analysis. If the third transcriber did not agree with one of the two original transcribers, the utterance was not included in the analysis.

Speech Analysis

A descriptive analysis of all the transcriptions was conducted to document type, frequency, and accuracy of consonant production. The subjects with severe speech production impairments may not have completed the entire word or sentence list, but analysis was conducted on any vocalization produced in response to the stimuli. The quantity of connected speech was highly variable among subjects. Any spontaneous speech was transcribed; however, 50 to 100 utterances were targeted for the sample. The samples were analyzed in two ways, by independent analysis and by relational analysis.

Independent Speech Analysis. An independent analysis documents the child's speech production inventory without comparison with the adult model (Stoel-Gammon and Dunn, 1985). This analysis provides information regarding the diversity of sound and syllable shapes used by the children when producing words. This measure describes the consonants that the child can produce, even though these sounds may be substituted for other sounds. An independent analysis was conducted to describe the type and frequency of consonant production for each subject. Consonant inventories were further categorized according to use in different positions of the word. A criterion of at least two productions of a consonant in each word position was required to include a consonant as present in a child's sound inventory.

Relational Speech Analysis. A relational analysis was conducted to compare the child's productions with the intended adult model. An error analysis was performed for each subject to examine the type of substitution or omission patterns used

TABLE 2 Percentage of Phenotypic Characteristics Exhibited by Subjects in the Velocardiofacial Syndrome (VCFS) and Comparison Groups

Characteristics	*VCFS Group, n = 13*	*Comparison Group, n = 8*
Cleft type		
Unilateral cleft lip and palate	8%	0%
Isolated cleft palate	8%	13%
Submucous cleft palate	38%	36%
Occult submucous cleft palate	31%	13%
Noncleft	15%	36%
Cardiac anomalies	46%	37%
Facial features		
Vertical maxillary excess	8%	13%
Hypotonic flaccid facies	77%	75%
Puffy eyelids	77%	0%
Overfolded helix	62%	0%
Small ears	46%	0%
Prominent nasal bridge	62%	50%
Bulbous nasal tip	69%	25%
Pinched alar base, narrow nostrils	77%	33%
Speech-language development history		
Severe hypernasality	63%	50%
Severe articulation impairment	15%	0%
Language impairment	77%	63%
Velopharyngeal insufficiency	100%	75%
Dysparaxia	85%	75%
High pitched voice	8%	13%
Hoarseness	8%	25%
Psychological and cognitive learning		
Learning disability	46%	50%
Borderline normal intelligence	15%	13%
Occasional mild mental retardation	23%	0%
Attention deficit hyperactivity disorder	23%	38%
Behavior problems in school	77%	38%
Physical development		
Motor delay	70%	63%
Feeding problems	23%	63%
Nasal regurgitation	54%	63%
Hearing history		
Frequent otitis media	70%	100%
Hearing loss-conductive	60%	88%
Hearing loss-sensorineural	0%	25%
PE Tubes	54%	75%
Intervention services		
Early intervention	38%	13%
Speech-language therapy	100%	88%
Hearing services	8%	13%
Other services: occupational therapy, physical therapy	15%	25%

Source: Information obtained by chart review utilizing the "Specialist fact sheet" (Shprintzen, 1998).

during the elicited and spontaneous speech samples. This analysis identified the type and frequency of errors that the subject produced. Two categories of errors, developmental and compensatory, were described. Developmental errors consisted of substitution and omission errors that are commonly observed in typically developing children and children with speech and language impairments (e.g., "tat" for "cat"). The number of developmental errors for the study population was compared with published norms described by Smit et al. (1990).

The type and frequency of compensatory substitution errors were analyzed for each subject. Compensatory errors are distinctive errors in place of articulation. For example, common compensatory articulation errors frequently observed for children with VP valving impairment are glottal stops and posterior nasal fricatives. For purposes of this study, the compensatory articulation types documented were those described by Trost-Cardamone (1997).

Another type of relational analysis is Percent Consonant Correct–Revised (PCC-R) described by Shriberg et al. (1997b). This analysis was used as a general measure of consonant accuracy in connected speech. The PCC-R was obtained by dividing the number of consonants correctly articulated, according to the adult model of production, by the total number of consonants used in the sample. In using the PCC-R, only consonant substitutions and deletions are calculated as incorrect. Distortions (e.g., nasal emission) are ignored with this analysis. This metric of articulation competence has been suggested to describe speech accuracy for children having diverse speech status as expected for the children in this study. Further, the PCC-R provides a rating for the severity level (e.g., normal, mild, moderate, or severe) of the child's speech production impairment. A criterion of at least five words was used as a minimum sample size on which to calculate PCC-R (Girolametto et al., 1997). Therefore, two subjects were omitted from the analysis of PCC-R because of severely restricted connected speech inventories.

Measures of Velopharyngeal Function

Multimethod evaluation of VP function was performed as part of the routine clinical evaluation. Perceptual and aerodynamic assessments were conducted by the first author for all subjects following a standard protocol described previously (D'Antonio et al., 1986). Fifteen of the 21 subjects underwent videotaped clinical endoscopic evaluation of VP function according to methods described previously (D'Antonio et al., 1988). Patients who did not exhibit substantial VP symptoms or who were noncompliant did not receive endoscopic evaluation at the time of the initial clinical assessment.

A composite rating of VP function was made for each patient on the basis of the collective results of the perceptual and instrumental assessments. A trichotomous categorization was used to describe VP function of each patient as demonstrating: complete VP closure, incomplete closure, or borderline closure. Videotapes of the endoscopic evaluations were reviewed by the first author and rated for several descriptive variables following suggestions for ratings of VP function made by an international working group (Golding-Kushner et al., 1990). These included degree of velar motion, degree of lateral wall motion, and presence of pulsing vessels. The reliability of such endoscopic ratings has been reported previously (D'Antonio, 1988).

Statistical Analysis

Responses are summarized by the means and SD. Differences between group means were assessed with analysis of variance (ANOVA), and multiple (pair-wise) comparisons of means were tested with the least significant difference (AN-

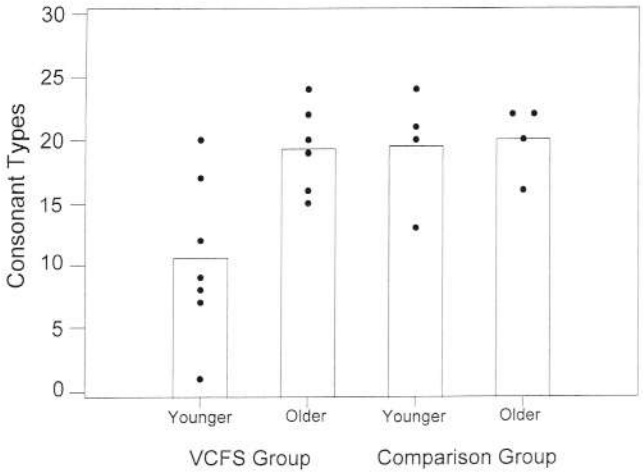

FIGURE 1 Number of consonant types for individual subjects for younger and older children in the velocardiofacial syndrome and comparison groups. Means are represented by the vertical rectangle.

OVA F-test is significant) or Tukey's procedure (ANOVA F-test is not significant). Mean ages of the children in the different study groups were not statistically different; therefore, adjustments for age were not employed when testing group means. The probability level of .05 or smaller was used to indicate statistical significance. Chi-square analysis was used to compare articulation development for the groups.

RESULTS

Independent Speech Analysis

As described previously, an independent analysis documents the child's speech production inventory without comparison to the adult model.

Consonant Inventory

The type and frequency of consonants used were calculated from the phonetic transcriptions for 20 of the 21 subjects. (Subject 9 in the older VCFS group was omitted because of an inadequate speech sample.) Patterns of sound use according to word initial or word final position showed no difference in the frequency of use between the positions; therefore, the results for the initial and final word positions were collapsed. Analysis of the sound inventories is shown in Figure 1. The mean number of consonant types used by the VCFS and comparison groups showed differences between the groups. The comparison group showed complete or near-complete consonant inventories for younger and older children. In contrast, the VCFS group showed fewer consonant types for the younger children with greater number of types for the older children. An ANOVA showed that the number of consonant types used by younger children in the VCFS group was statistically less than the number used by the older children in the VCFS group, the younger children in the comparison group, and the older children in the comparison group ($F = 5.45$, $p < .05$) with the Tukey procedure performed after ANOVA.

Table 3 shows the specific sound inventories produced by each subject listed in order of youngest to oldest subject for both the VCFS and comparison groups taken from imitated word lists and recognized word attempts. The circles represent use of a sound at least twice in the elicited or spontaneous speech sample according to the criteria described by Stoel-Gammon and Dunn (1985). These inventories represent the optimal sound repertoire that the child can produce in any context.

Manner of consonant production refers to how the air stream is modulated by the articulators (e.g., stops, fricatives, nasals, etc.). The manner of consonant production was analyzed for individual subjects. All children, except subjects 5 and 9, produced consonants in each manner of production category, but the number of sounds in each category varied. A comparison of the younger children in the VCFS group revealed fewer consonant types in every manner category. Additionally, there appeared to be a preference for voiceless consonants within the broader categories. Comparison of the older groups showed similar numbers of consonants within the manner categories with the exception of the affricate category.

Place of consonant articulation refers to the location in the vocal tract in which the primary modulation of the air stream occurs (e.g., bilabial, labiodental, glottal, etc.). When place of articulation was examined within manner categories, four of the seven younger children in the VCFS group showed restrictions in place of articulation features. These children used sounds made at the extremes of the vocal tract (i.e., front and glottal). In contrast, the older children in the VCFS group appeared to demonstrate all places of articulation. Inspection of Table 3 shows that younger and older children in the comparison group demonstrated nearly complete sound inventories.

Frequency of Consonant Use

When examining sound inventories, the number of productions or general talkativeness of the child will influence the absolute number of consonants used. However, in this study, the VCFS and comparison groups had equivalent numbers of utterances making comparisons between the groups possible. The younger children in the VCFS group demonstrated the lowest frequency of consonant use ($\bar{X} = 98.5$, SD = 67.5 consonants) followed by the younger comparison group ($\bar{X} = 141$, SD = 63.0 consonants), the older VCFS group ($\bar{X} = 159$, SD = 44.0 consonants) and the older comparison group ($\bar{X} = 177$, SD = 37.8 consonants). Although the frequencies of consonant use among the groups varied, statistical analysis showed no significant difference among the groups for the frequency of consonants used ($p = .13$).

Relational Speech Analysis

A relational analysis compares the child's speech productions to the intended adult model.

TABLE 3 Consonant Sound Inventories for Individual Subjects in the Velocardiofacial Syndrome (VCES) and Comparison Groups, Listed from Youngest to Oldest Subject Each Group Taken from Imitated Word Lists and Recognized Word Attempts. Circles Represent the Use of a Sound At Least Twice

	PHONEMES																												
	STOPS						FRICATIVES								AFFRICATES		GLIDES		LATERALS		NASALS			COMPENSATORY ERRORS			
SUBJECT	p	b	t	d	k	g	f	v	s	z	ʃ	ʒ	ð	θ	h	ʧ	ʤ	w	j	l	r	m	n	ŋ	ʔ	ɾ	ɬ	Δ
VCFS-younger 1	○	○		○	○	○	○		○						○	○		○	○	○	○	○	○		○			○
2	○						○		○						○	○					○	○	○		○			
3	○						○								○			○	○	○	○	○			○			
4	○												○		○			○				○	○		○			
5																						○						
6	○	○	○	○	○	○	○	○	○	○	○				○	○	○	○		○	○	○	○	○	○			
7	○		○		○		○	○	○		○				○					○		○	○					
VCFS-older 8	○	○	○	○	○	○	○	○	○	○	○				○			○	○	○	○	○	○					
9																												
10	○	○		○	○		○		○				○	○	○			○		○	○	○	○		○			
11	○	○	○	○	○	○	○	○	○	○	○			○	○	○	○	○	○	○	○	○	○		○			
12	○	○	○	○	○	○	○		○	○	○			○	○	○		○	○	○	○	○	○		○			
13	○	○	○	○	○	○	○	○	○	○	○		○	○	○	○	○	○	○	○	○	○	○	○	○			
Comparison -younger 1	○	○	○	○		○	○		○	○	○		○		○		○	○	○	○		○	○	○	○	○		
2	○	○	○	○	○	○	○	○	○	○	○			○	○	○	○	○	○	○	○	○		○	○	○		
3	○	○	○	○	○	○	○	○	○	○	○			○	○	○	○	○	○	○	○	○	○	○	○			
4	○	○	○	○	○	○	○	○	○	○	○				○	○	○	○	○	○	○	○	○		○			
Comparison -older 5	○	○	○	○	○	○	○	○	○	○	○		○	○	○	○		○	○	○	○	○	○	○	○			
6	○	○	○		○		○	○						○	○			○	○		○	○	○		○		○	
7	○	○	○	○	○	○	○	○	○	○	○		○		○	○	○	○	○		○	○	○		○			
8	○	○	○	○	○	○	○	○	○	○	○				○	○	○		○	○	○	○	○		○			

Error Analysis and Normative Comparisons

There were no differences in the types of developmental articulation errors displayed by the VCFS and comparison groups. Younger children in the VCFS group displayed an average of 10.7 types of developmental articulation errors, and younger children in the comparison group demonstrated an average of 10 error types. Similarly, older children in the VCFS group displayed 7.8 developmental error types, and their counterparts in the VCFS group showed 8.0 error types.

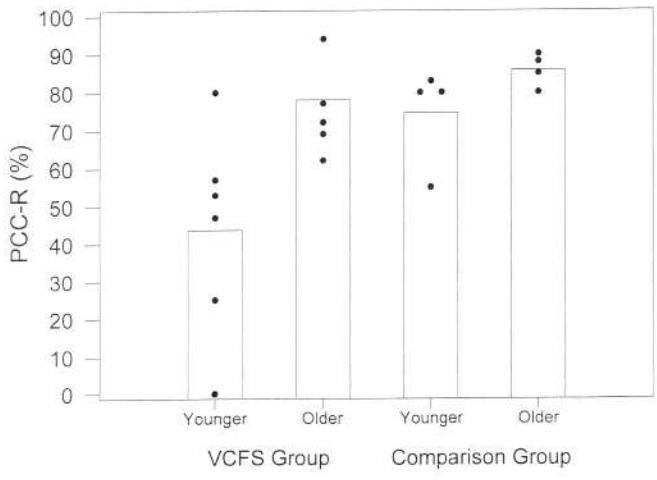

FIGURE 2 Percent Consonant Correct–Revised (PCC-R) for individual subjects for younger and older children in the velocardiofacial syndrome and comparison groups. Means are represented by the vertical rectangle.

It is important to recall that the groups being compared are both clinical populations with identifiable speech production disorders. The present study did not use a normal control group. However, the performance of the subjects from this study can be compared with normative data reported by Smit et al. (1990) that provides the age at which 75% of normally developing children acquire accurate consonant production. A significantly lower number of children in the young VCFS group achieved age-appropriate articulation development, compared with the other groups (chi-square $p = .05$). Only 17% of the younger children in the VCFS group reached the criteria for normal consonant production for their individual age as reported by Smit et al., and 50% of the older children in the VCFS group reached normalized consonant articulation. In contrast, younger children in the comparison group presented with 100% normalization for their individual ages and 50% of the older comparison group reached the criteria.

Percent Consonant Correct–Revised

The PCC-R was calculated to provide a measure of consonant accuracy and was calculated for all children who used

more than five words. Figure 2 shows the PCC-R for the younger and older children in the VCFS and comparison groups. The younger children in the VCFS group exhibited a significantly lower PCC-R from the older children in the VCFS group or from both younger and older children in the comparison group ($F = 6.88$, $p = < .05$) with Tukey procedure performed after ANOVA.

Severity descriptors were assigned to the PCC-R calculations as described by Shriberg et al. (1997b). Based on the average PCC-R calculations, results showed the younger children in the VCFS group demonstrated a severe impairment, and children in the older VCFS group and the children in the younger comparison group presented with a mild severity of impairment. The children in the older comparison group showed no impairment.

Compensatory Articulation Errors.

Analysis of the use of compensatory articulation errors showed differences between the groups. The articulation of children with VCFS has been characterized as having a high occurrence of compensatory articulation errors, particularly glottal stops that are frequently substituted for whole classes of consonant types. The predominant compensatory articulation error for all groups was glottal stops with infrequent use of the pharyngeal stop, pharyngeal fricative, and posterior nasal fricative. The compensatory articulation error type used predominantly by the VCFS group was the glottal stop substitution, and subjects in the comparison group were more likely to use other types of consonant substitutions in addition to the use of glottal stops.

The frequency of glottal stop use was calculated for each child as a "percentage of glottals" defined as the number of glottal stop substitutions divided by total oral consonants used. For this analysis, the total speech production sample was used to provide a larger corpus of utterances. Table 4 shows the percent of glottal stop consonants used by the younger and older subjects in the VCFS and comparison groups. Although the younger subjects in the VCFS group used the highest percentage of glottal stops, results of an ANOVA showed no statistically significant difference among the groups for percentage of glottals.

TABLE 4 Percent Glottal Stop Consonants Used in All Vocalizations for Children in the Velocardiofacial Syndrome VCFS and Comparison Groups

Group	*Subjects*	*Age, y, mo.*	*Glottal Consonants (%)*
1	VCFS (younger)		
	1	3, 4	29/157 18
	2	4, 4	16/40 40
	3	4, 8	81/96 84
	4	5, 6	21/54 38
	5	5, 8	0/0 —
	6	5, 11	14/120 12
	7	6, 1	7/55 13
			Mean 27.8
2	VCFS (older)		
	8	7, 3	24/131 18
	9	7, 7	25/89 28
	10	7, 6	21/81 26
	11	9, 9	16/131 12
	12	10, 2	43/159 27
	13	11, 0	10/152 6
			Mean 19.5
3	Comparison (younger)		
	1	5, 2	70/156 44
	2	6, 4	20/182 11
	3	6, 7	18/131 14
	4	6, 7	11/104 11
			Mean 20.0
4	Comparison (older)		
	5	7, 7	31/150 21
	6	8, 1	20/62 32
	7	8, 6	20/169 12
	8	9, 6	6/73 8
			Mean 18.3

Measures of Velopharyngeal Function

Table 5 shows the subjects in the VCFS and comparison groups and their composite ratings of VP function (i.e., adequate, inadequate, or borderline VP closure). Only one child in the older VCFS group and one in the younger comparison group demonstrated adequate VP closure. The remainder of the subjects in the VCFS group (n = 8/13, 62%) had inadequate VP closure or borderline closure (n = 4/13, 30%). A smaller number of the children in the comparison group demonstrated inadequate VP closure (n = 2/8, 25%), and the predominant pattern for the comparison group was borderline closure (n = 5/8, 62%). There was no statistically significant difference between the VCFS and the comparison groups in the distribution among the VP function categories.

Review of the endoscopic videotapes showed that two of the nine subjects in the VCFS group showed no evidence of any velar elevation (subjects 5 and 7 in the VCFS group), and no subjects in the comparison group demonstrated a complete absence of motion. Similarly, four of nine subjects in the VCFS group demonstrated no observable lateral wall motion, and only one subject in the comparison group presented with an absence of lateral wall movement. Finally, 50% of the subjects in the VCFS group presented with endoscopic evidence of abnormal vasculature, and no subjects in the comparison group demonstrated this feature.

Relationship Between Velopharyngeal Function and Consonant Production

The relationship between VP function and several measures of consonant production is shown in Table 5. There appeared to be a complex relationship between subject groups, VP function category, and the consonant variables. To facilitate interpretation of these data and to provide larger sample sizes in the comparison groups, some of the variables were collapsed for post hoc analysis. Figure 3 displays the data with the adequate and borderline VP closure categories collapsed. Additionally, the preceding results throughout this study consis-

TABLE 5 Relationship Among Cleft Type, Velopharyngeal (VP) Function, and Articulation Variables for Subjects in the Velocardiofacial Syndrome (VCFS) and Comparison Groups

Group	*Subject*	*Cleft Type**	*VP Function*	*Consonant Types*	*PCC–R**	*Severity Rating*	*Glottals (%)*
1	VCFS (younger)						
	1	CP	borderline	17	57	moderate-severe	18
	2	OSMCP	inadequate	8	0	severe	40
	3	SMCP	inadequate	9	47	severe	84
	4	UCLP	inadequate	7	25	severe	38
	5	OSMCP	inadequate	1	—		—
	6	NC	borderline	20	80	mild-moderate	12
	7	SMCP	inadequate	12	53	moderate-severe	13
	Mean			10.6	43.7		27.8
2	VCFS (older)						
	8	SMCP	inadequate	19	77	mild-moderate	18
	9	SMCP	inadequate	15	62	moderate-severe	28
	10	VPDYS	adequate	16	69	mild-moderate	26
	11	OSMCP	borderline	22	94	wnl*	12
	12	SMCP	inadequate	20	72	mild-moderate	27
	13	OSMCP	borderline	24	94	wnl	6
	Mean			19.3	78.0		19.5
3	Comparison (younger)						
	1	OSMCP	borderline	20	55	mild-moderate	44
	2	NC	borderline	24	80	mild-moderate	11
	3	CP	borderline	13	80	mild-moderate	14
	4	VPDYS	adequate	21	83	mild-moderate	11
	Mean			19.5	74.5		20.0
4	Comparison (older)						
	5	SMCP	borderline	22	85	wnl	21
	6	OSMCP	inadequate	16	80	mild-moderate	32
	7	SMCP	inadequate	22	90	wnl	12
	8	SMCP	borderline	20	88	wnl	8
	Mean			20.0	85.8		18.3

* CP = cleft of secondary palate; OSMCP = occult submucus cleft palate; SMCP = submucous cleft palate; UCLP = unilateral cleft lip and palate; VPDYS = velopharyngeal dysproportion; NC = noncleft; PCC–R = percent consonant correct–revised; wnl = within normal limits.

tently showed that the younger children in the VCFS group differed from the older children with VCFS and the younger and older children in the comparison group. Therefore, for this analysis, the younger and older children with VCFS were displayed separately, and the younger and older children in the comparison group were combined into one group.

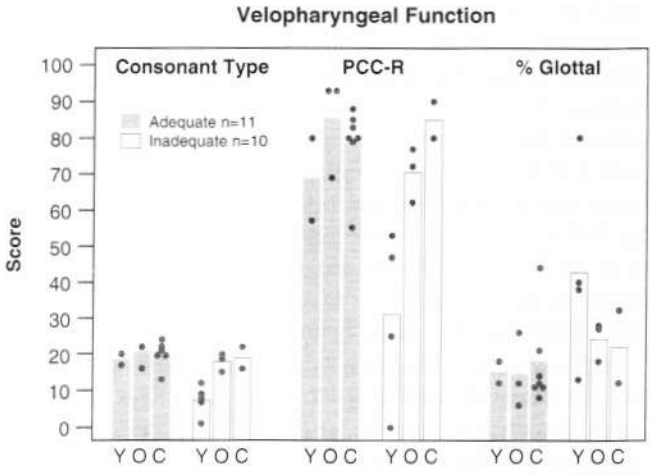

FIGURE 3 Relationship between velopharyngeal function and three measures of consonant production for younger children in the velocardiofacial syndrome (VCFS) group (Y), older children in the VCFS group (O), and children in the comparison group (C). Composite ratings of velopharyngeal function are collapsed into two categories, adequate and borderline and inadequate. Scores represented include number of consonant types, Percent Consonant Correct–Revised (PCC-R), and percent glottal stop substitutions for oral consonants produced (% glottal).

Inspection of Figure 3 demonstrates when VP closure was adequate or borderline, there were no differences among the younger children with VCFS, older children with VCFS, or the comparison group for any of the consonant variables. However, when VP closure was inadequate, younger children with VCFS demonstrated significantly fewer consonant types ($p < .05$) and significantly lower PCC-R than older children with VCFS ($p < .05$) or children in the comparison group ($p < .05$). There were no differences between the groups in the percentage of glottal stop use.

Discussion

The present study is descriptive in nature and assesses a relatively small number of children. However, it differs from previous reports in a manner that is important for future investigations. There is vast literature that has established accepted protocols and methodologies for describing speech and language development in typically developing children and clinical populations. These metrics tend to differ from the information routinely gathered in clinical evaluations of speech and language. Because most of the data available regarding communication development in children with VCFS have been collected using a clinical paradigm, these accepted research methodologies have not been employed in the collection, anal-

ysis, or reporting of data. This has made comparison between the VCFS population and the research data pertaining to other clinical populations difficult, if not impossible. The value of the present study is that it utilizes a method for analyzing speech patterns in children with VCFS employing standard, accepted research metrics adapted for clinical practice to describe the speech characteristics of a sample of children who were originally evaluated during routine clinical practice.

This study addressed two specific research questions that have important implications for future studies regarding the profile and course of communication impairment displayed by children with VCFS. The first question asked whether the speech production characteristics of children with VCFS differ from children with speech impairment who also demonstrate some of the phenotypic characteristics of VCFS but who do not have the syndrome. Information regarding the similarities and differences between these groups would provide valuable insights into the broader question of whether the profile of communication impairments displayed by children with VCFS is unique to the syndrome. The second specific question asked whether younger children with VCFS demonstrate the same pattern of speech impairment as older children with VCFS. Information regarding the speech patterns of children with VCFS at different developmental levels would provide insights into hypotheses concerning the developmental course of communication impairment associated with VCFS. Data from this study provided answers to the specific research questions that were asked and provided important information necessary for further exploration of the broader research issues.

Specific Research Questions

Results of this study showed that the children with VCFS demonstrated several speech production patterns that were different from speech patterns of a comparison group of children with some of the features of VCFS who do not have the syndrome. The features that differentiated the groups were most prominent in the younger children. Although older children in the VCFS group were not age appropriate with respect to speech development, they were similar to the comparison group of children with speech deficits who did not have a diagnosis of VCFS.

The younger children with VCFS were qualitatively and quantitatively different from older children with VCFS or children in the comparison group. Inspection of the independent speech analysis showed that younger children with VCFS had a smaller repertoire of consonant types as evidenced by their reduced consonant inventories. The younger children with VCFS were capable of producing consonants with all manner features (such as stops, fricatives, etc.); however, the number of consonant types within each manner category was lower for the young children with VCFS. Additionally, this group appeared to demonstrate a preference for voiceless consonants regardless of the place or manner of articulation. The younger children with VCFS also demonstrated the lowest frequency of consonant use compared with the other groups.

Results of the relational speech analysis showed a similar pattern to that observed in the independent analysis. When comparing the children in this study with normative data, far fewer children in the younger VCFS group demonstrated age-appropriate consonant production than in the older VCFS group or either the younger or older children in the comparison group. Furthermore, results of the relational speech analysis showed no differences in the types of developmental articulation errors used by the children in the VCFS and comparison groups. On the other hand, analysis of compensatory articulation errors showed that all children produced compensatory articulation errors. However, the compensatory articulation error type used predominantly by children with VCFS was the glottal stop, and subjects in the comparison group were more likely to use other types of consonant substitutions in addition to the glottal stop. Comparison among the groups showed that the younger children with VCFS demonstrated the highest use of glottal stop errors, compared with the other three groups. The younger children in the VCFS group also demonstrated lower levels of consonant accuracy (as measured by PCC-R and compared with age matched norms), compared with children in the older VCFS group or children in the comparison group.

Ninety percent of the children in this study demonstrated evidence of VP inadequacy. This high rate of VP symptoms is related to the ascertainment method, whereby subjects for this study were referred for evaluation of speech and VP function following screening by a Craniofacial team. There did appear to be a trend for children with VCFS to demonstrate a higher occurrence of VPI than children without VCFS; however, this difference was not statistically significant. However, there was an important difference between the groups that was clinically important. No children in the comparison group demonstrated a complete absence of velar motion. On the other hand, two of the nine subjects in the VCFS group who had endoscopic evaluations showed no velar activity for speech, and four of the nine demonstrated no observable lateral wall motion. This finding is further support that there are differences in the speech production mechanisms of some children with VCFS that are not commonly observed in children with some similar speech patterns but who do not have a diagnosis of VCFS. Similarly, in this population, 50% of the children with VCFS who had endoscopic evaluations had evidence of abnormal pulsations on the posterior pharyngeal wall. Again, this finding may be relatively unique to the syndrome.

The results from this study provide new information regarding the nature and origin of the speech production characteristics observed in children with VCFS. A common clinical assumption is that the unusual speech production patterns displayed by children with VCFS, particularly the high occurrence of glottal stop errors, were related in a direct causal manner to the presence of VPI. The findings from this study indicated that this relationship may be more complex than has been suggested previously and age may be a critical variable.

This finding that the relationship between VP function and speech patterns observed in children with VCFS is more com-

plex than has been described previously is supported by an earlier study that compared the speech development of children with VCFS with children with palatal clefting (Scherer et al., 1999). Data from that study showed that the speech development of infants and young children with VCFS differed from and showed more impairment than comparison groups of children with cleft lip and palate or isolated cleft palate, many of whom demonstrated similar magnitude of VP symptoms.

Broader Theoretical Questions

The results from this study provide new information concerning the hypothesis that children with VCFS present with a unique profile of speech impairment that is distinct from normal developing children or children with other patterns of speech and language impairment. The present study provided data to support this hypothesis. Results showed that young children with VCFS demonstrated some speech production patterns that differ from normal developing children (as evidenced by comparison with normative data) and from older children with VCFS and children with communication impairment with some features of VCFS who do not have the syndrome.

The data from this study also contributed information concerning the developmental course of communication impairment in children with VCFS. The study by Scherer et al. (1999) raised questions regarding the developmental course of the speech impairments observed in children with VCFS. Data from that study documented severe speech and language impairments in young children with VCFS that could be identified as early as 6 months of age and increased in severity, becoming distinct from normal developing children and children with clefts by 12 to 18 months of age. However, the course of these impairments beyond 30 months of age was unclear. Data presented by Solot et al. (1998) suggested that the speech production impairments observed in young children with VCFS improved greatly by school age. Similarly, Shprintzen has suggested a rapid "catch-up" period between 3 and 5 years of age (personal communication). Data from the present study interpreted collectively with the data presented by Scherer et al. (1999) suggest a developmental progression of improving speech production. Specifically, the first study documented severe speech production impairment for young children under 30 months. Results from the present study documented severe speech impairment in children 3 to 7 years old with VCFS and mild impairments in 7- to 10-year-olds with VCFS. However, for the young children with VCFS in the present study, although the speech production impairments were clearly severe, they were less severe than those observed for the children under 30 months of age as documented previously. These findings may suggest a developmental progression of gradual improvement of speech production. It is unclear whether this developmental progression is mirrored on overall cognitive development or motor development or follows a pattern of catch up that is specific to the speech production system.

Directions for Future Research

The present study contributes information valuable for developing hypotheses and new research questions regarding communication impairment in children with VCFS. The data support the hypothesis that some children with VCFS may present with a distinct profile of communication impairment. Additionally, the data suggest there may be a characteristic developmental progression of communication impairment in some children with VCFS with a period of rapid catch-up.

However, the present study was conducted to provide pilot data to facilitate hypothesis development and to assist in identifying salient variables for future study. Several methodologic limitations of this study may preclude generalization of the data. For example, the gender distribution of the current study population was predominantly female; however, since speech and language development is generally accelerated in girls, compared with male counterparts, the high number of females in this study would not be expected to overemphasize the level of delay. Another procedural issue that may preclude generalization of the data is that the subjects from this study were all selected through referral from a craniofacial team, and there may have been some ascertainment bias whereby children with the most severe communication impairment were referred at a younger age. Although the age distribution of the younger children in the VCFS and comparison groups did not differ from one another statistically, there were some differences in the distribution that may have affected the results. In spite of the strong findings that younger children did poorer across speech production measures, there was only one child under 4 years of age (in the VCFS group). Therefore, it is possible that the severity of the impairments may have been underestimated because of this underrepresentation of 3-year-old children. On the other hand, there were no children under 5 years in the comparison group, and this may have skewed the results such that the more severe disorders in the VCFS group were related to the young age of the children in that group. However, results of the various relational analyses in which performance of the subjects was compared with age-matched normative data demonstrate that the young children with VCFS differed more from the norms than young children in the comparison group.

Another important methodologic limitation of this study is the lack of detailed quantitative information regarding the developmental status of the subjects. Children entered this study through clinical evaluation and had a variety of physiological and educational assessments that were not comparable. Therefore, the impact of developmental level on the findings in this study is unknown. It is important to note that the present data support results of clinical audits of larger populations of children with VCFS (Solot, 2000; Perrson and Oskarsdottir, 2000). However, in the future, studies of large populations of children with VCFS are needed in which the speech data are analyzed in detail in relationship to developmental data. Another limitation of this study is that the speech samples in this study were collected during a routine clinical evaluation, and no attempt was made to standardize the spontaneous speech sample

or the length of the sample. Finally, the current study did not assess measures of intelligibility or naturalness. Future studies should be designed to address the contribution of the various speech characteristics to the poor intelligibility often demonstrated by children with VCFS.

In spite of the limitations of the existing data set, the results support the need for prospective longitudinal studies of larger samples of children with VCFS and appropriate controls. Results of such studies will provide the foundation for developing treatment plans based on the individual profiles of impairment and stage in the developmental progression of the disorder. Once treatment plans have been developed, intervention studies can be designed and conducted.

Clinical Implications

It is a common clinical observation that the speech characteristics of many children with VCFS differ in a variety of ways from normally developing children and children with other types of communication impairment. Scherer et al. (1999) demonstrated that the speech and language development of young children with VCFS differs from the developmental patterns of children with cleft lip or cleft palate. Similarly, the results of the present study demonstrate that the speech patterns of young children with VCFS differ not only from normal developing children and children with cleft palate but also from children with features similar to those found in VCFS but who do not have the syndrome. Interpreted collectively, the results of these studies suggest there may be a syndrome specific communication profile demonstrated by some children with VCFS. These findings would suggest that in clinical practice when a child presents with speech and language characteristics similar to those described, a diagnosis of VCFS should be considered. In fact, in some children the speech patterns may be the first feature identified that leads to accurate diagnosis.

In spite of the data demonstrating the severity and the characteristic communication patterns associated with VCFS, there has been little attempt to take these patterns into consideration when planning for evaluation and intervention. This study provided an in-depth description of the speech production characteristics of a small group of children with VCFS. The present data interpreted with clinical experience suggest that the impairments presented by many children with VCFS are complex. This complexity, therefore, warrants (and in fact, requires) in-depth evaluation strategies that may not be routine in many settings. For example, in the present study, an analysis of differences between articulation skills in single words and connected speech was not reported due to the unequal number of utterances in the spontaneous speech sample and the limited number of elicited single words. However, there appeared to be some differences between speech skills in these two conditions. For example, the data presented in Table 3 concerning consonant inventory were derived from imitated word lists and recognized word attempts in structured contents. Three children did not demonstrate the use of glottal stop articulation in these contexts. However, analysis of speech as shown in Table 4 revealed glottal stop articulation for all subjects except one child whose only word attempt was "mom." This illustrates an important point that should be considered clinically. Children with VCFS are frequently tested using a standard articulation test in which they may show the capability of producing all or most consonants accurately at the single word level but substitute glottal stops for whole classes of consonants in connected speech. A discrepancy between articulation skills for single-word production and connected speech can have far-reaching implications. For example, one subject in this study presented with pervasive glottal stop substitutions in connected speech but demonstrated the capability of producing all consonants accurately during a standard articulation test. The Individualized Education Plan developed by the child's school district reported "no articulation errors" and judged her to be ineligible for further speech therapy based on assessment of articulation in elicited single words. Such cases point to the need for sampling of articulation in both single word and connected speech contexts. Furthermore, in young children with VCFS, it is important to augment traditional assessments of articulation. Recall that the most commonly utilized measures of articulation compare the child's productions with the adult model. However, it is valuable to examine the child's individual speech sound system independent of the adult model. This is particularly important when a young child is highly unintelligible and the average listener is not recognizing the sound substitution patterns. Furthermore, accurate description of an individual child's sound system provides critical data for developing optimal treatment targets.

Although the speech production deficits demonstrated by children with VCFS have an obvious impact on communication, they may also have more far-reaching deleterious effects. Children with VCFS have a variety of psychoeducational issues and are more likely than other children with speech production impairment to receive large batteries of tests at an early age to assess cognitive, psychological, and educational functioning. The severe communication impairment, particularly the high percentage of glottal stop consonants resulting in severe intelligibility impairment, makes such testing difficult. Often appropriate responses are not recognized by an unfamiliar listener and the child is not credited with a correct response. Therefore, care should be exercised in conducting such tests and interpreting the results.

The data from the present study provide a detailed analysis of the speech characteristics of a sample of children with VCFS. Such studies are requisite steps in developing intervention studies and subsequent treatment paradigms. However, to date, no treatment studies have been published, and there is little consensus regarding the appropriate course of treatment for the speech and language impairments associated with VCFS. Further research is needed to elucidate the profiles and developmental course of communication deficits in children with VCFS.

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