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Early Speech and Language Development in Children With Velocardiofacial Syndrome

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Speech–language impairment is one of the most common clinical features in velocardiofacial syndrome (VCFS). This report describes the speech and language development of four children with VCFS studied longitudinally from 6 to 30 months of age and compares their performance with three groups of children: (1) normally developing children, (2) children with cleft lip and palate, and (3) children with isolated cleft palate. The data show that young children with VCFS show a receptive-expressive language impairment from the onset of language. Further, speech and expressive language development were severely delayed beyond a level predicted by their other developmental or receptive language performance. The children with VCFS showed severe limitations in speech sound inventories and early vocabulary development that far exceeded those shown by the children with cleft lip and palate and children with isolated cleft palate. This study indicates that young children with VCFS emerge from a critical speech and language learning period with severe limitations in their communicative abilities. Further studies are required to describe the later course of these early speech and language impairments and to explore the relationship to learning disabilities described for older children with VCFS. Am. J. Med. Genet. (Neuropsychiatr. Genet.) 88: 714–723, 1999. © 1999 Wiley-Liss, Inc.

KEY WORDS: velocardiofacial syndrome; 22q11 deletion; speech–language impairment; cleft palate

INTRODUCTION

Velocardiofacial syndrome (VCFS) is a multiple anomaly disorder first described by Shprintzen et al. in 1978. Many clinical features have been described, with the most common findings including cleft palate (overt, submucous, or occult submucous), conotruncal heart anomalies, characteristic facies, and learning disabilities. Golding-Kushner et al. [1985] provided a description of the language, academic, and psychological profiles of 26 patients with VCFS ranging in age from 3 to 18 years. In their study, speech and language impairments included reduced understanding and reduced use of vocabulary, syntax, and abstract reasoning in communicative situations. Velopharyngeal insufficiency (VPI) was found in all cases. A recent study of 181 children with VCFS showed an even broader range of communication and developmental impairment [McDonld-McGinn et al., 1997]. Solot et al. [1998], studying a subgroup of the same sample, reported both receptive and expressive language impairments with expressive language being more severely impaired than receptive language. Early speech and language development was characterized as delayed in onset with most children nonverbal at age 2 years. Language impairments continued to be a prominent feature of the developmental profile of children in the preschool and school aged periods.

It is accepted clinically that the speech development of children with VCFS differs in many ways from normally developing children [Golding-Kushner et al., 1985; Nayak and Sells, 1998; Solot et al., 1998]. For example, there is an unusually high occurrence of glottal stop substitutions in children with VCFS. (A glottal stop is a sound made by stopping air with the vocal folds instead of in the oral cavity.) Typically, glottal stops are substituted for the whole class of stop consonants (p, b, t, d, k, g) often rendering speech unintelligible. There are some similarities to the speech development of children with cleft lip and/or palate. These include characteristics such as VPI, use of compensatory speech sounds (e.g., glottal stops), early language impairment, and some learning disabilities [Broder et

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TABLE I. Descriptive Data for the Subjects*

Subject	Cleft type	Gender	Age at palate repair	Deletion	Onset of SL therapy
1	NC, DP	M	—	de novo	18 m
2	CP	M	12 m	de novo	26 m
3	SMCP, DP	F	—	Familial (maternal)	12 m
4	CP	F	13 m	Familial (maternal)	20 m

*CP = cleft of the secondary palate; SMCP = submucous cleft; NC = noncleft; SL = speech–language; m = months of age; DP = deep pharynx.

al., 1998; Richman and Eliason, 1988; Scherer and D'Antonio, 1995]. However, it remains unclear whether the speech and language patterns of young children with VCFS are similar in type, prevalence, and severity when compared with children with palatal clefting.

Also, even though speech and language impairment is nearly universal in children with VCFS, little is known about the development of these skills between birth and 3 years of age. With the identification of the 22q11.2 deletion in patients with VCFS, larger numbers of individuals are being diagnosed with the syndrome. Concurrently, there is an emphasis on early intervention for children with developmental disabilities. It is therefore important to understand the earliest developmental patterns of children with VCFS.

This paper describes a detailed and comprehensive assessment of the speech and language development in four children with VCFS from 6 to 30 months of age and contrasts this development with three comparison groups—normally developing children, children with cleft lip and palate, and children with isolated cleft palate.

MATERIALS AND METHODS

Subjects

Children with VCFS. Four children with VCFS were studied. Table I shows the cleft type, gender, age at palate repair, intervention history, and whether the deletion was familial or de novo. Two of the children had overt clefts of the palate, one child had a submucous cleft with a deep pharynx, and one child did not have an identified cleft but had a deep pharynx. For the children with VCFS, Subjects 1 and 2 were enrolled in speech and language therapy at 18 and 26 months of age, respectively. Subjects 3 and 4 speech and language therapy between 12 and 20 months of age.

The diagnosis of VCFS was confirmed by fluorescent in situ hybridization (FISH) showing a deletion of 22q11.2. Two of the children (Subjects 1 and 2) lived in intact families with their biologic parents. For these children neither parent was deleted. Subjects 3 and 4 lived in an extended family with their mothers who were also affected and with the maternal grandparents who did not have any of the characteristics of VCFS.

Comparison Groups. Three groups of children were followed with the same protocol as the children with VCFS and the data for the groups are discussed here for purposes of comparison. These groups included normally developing children and children with cleft lip and palate. Previous studies have demonstrated a relationship between cleft type and the presence of language and learning disorders [Jones, 1988; Richman and Elaison, 1995; Scherer and D'Antonio, 1997]. Therefore, for this study two groups of children with clefts were used for comparison purposes; children with cleft lip and palate (CLP) and children with isolated cleft palate (ICP). The children with CLP were free from syndromes as determined by a geneticist. Table II shows the description of the children in the comparison groups. Twenty-three children were followed: eight typically developing children, eight children with CLP, and seven children with ICP.

The children with CLP, ICP, and VCFS were matched to the children in the normal group based on socioeconomic status and gender. The groups were controlled for timing of palate repair. All of the children in the cleft and VCFS groups were receiving some form of early developmental treatment during the study.

Procedures

Evaluations were performed at 6, 12, 18, 24, and 30 months of age for all children. The children were evaluated in their homes with their mother and caretakers present. A single evaluator performed all the developmental, language, and speech testing.

Hearing

Hearing screenings and tympanometry were performed at each visit by a certified, licensed audiologist. All the children with clefts had pressure equalization

TABLE II. Descriptive Data for the Comparison Subjects*

Subject	Gender	Age at palate repair	BSID-2 Mental	BSID-2 Motor
Normal				
1	M	NA	97	108
2	F	NA	106	95
3	M	NA	90	110
4	M	NA	130	118
5	F	NA	108	108
6	M	NA	105	115
7	M	NA	112	105
8	F	NA	102	107
CLP				
1	M	15 M	96	98
2	M	7 M	112	101
3	M	12 M	97	115
4	F	18 M	102	115
5	M	15 M	112	108
6	M	11 M	110	100
7	M	10 M	92	96
8	F	12 M	107	101
ICP				
1	M	10 M	109	100
2	M	12 M	74	80
3	F	12 M	71	100
4	M	12 M	76	84
5	F	16 M	83	90
6	F	15 M	60	75
7	M	12 M	65	60

*CLP = cleft lip and palate; ICP = isolated cleft palate; NA = not applicable; M = months of age.

tubes placed at the time of palate surgery. The child with VCFS without a cleft had tubes placed between the 24 and 30 month evaluations. All the children had failed tympanometry and some hearing acuity screenings prior to tube placement; however, all passed hearing screenings following tube placement.

Developmental Measures. All the children received the mental and motor scales of the Bayley Scales of Infant Development-2 (BSID) at 6, 18, and 30 months of age. Young children are often given general measures of development to monitor their performance through the first 3 years of life. These general measures sample development from many domains (i.e., visual-motor, receptive language, expressive language, speech sounds, feeding, social, fine and gross motor). Only after 3 years of age are these developmental domains assessed separately. Of these domains that are tested in early development, language milestones are a prominent feature of development between 18 months and 3 years of age. If the goal is to identify a child's cognitive abilities apart from speech and language impairment, then the language items should be separated from the nonlanguage items. Because we suspect that children with VCFS have language delays, this was the methodology employed in this study. The BSID provides an item analysis for language, cognitive, social, and motor skills that were used to separate language and nonlanguage performance.

When interpreting the scores on the motor and mental scales of the BSID, scores that fall between 85–115 are considered to be in the normal range. Scores from 70–85 are considered to be in the borderline range, and scores below 70 are indicative of delays.

General Measures of Language Development. Receptive and expressive language performance were measured using the *Sequenced Inventory of Communicative Development-R* (SICD) [Hedrick et al., 1984] at all assessments. The SICD is a standardized measure of language development for children 4 months to 4½ years of age, yielding age equivalent scores. It has known validity and is widely used in research on speech and language impairments. This measure assesses receptive and expressive language development across multiple language domains including vocabulary, pragmatics, grammar, and phonology.

Language Sample. General tests of language development sample a variety of language parameters across ages, but not in detail. Thus only a global picture of the child's language performance can be obtained from the SICD. The second method of language assessment examines the child's use of language in a communicative context called a language sample. This method provides a more typical view of the child's ability to use language routinely. Language sample analysis was performed using a 30-min videotape sample of communicative interaction between the child, caregivers, and examiner. The strength of a language sample analysis is that it assesses language use in a natural conversation rather than a "testing" situation, which requires that the child respond to specific items. For young children this method is preferred because it reflects the child's use of language within typical contexts.

Methodologically, a sample of 30 min was selected to give children sufficient opportunity to use language. This sample length is in concert with established literature in language assessment [Miller, 1981]. The language sample analysis was performed on the samples obtained at the 12- through 30-month assessments using the Systematic Analysis of Language Transcripts [Miller and Chapman, 1996]. In preparing the language sample for analysis, any vocalization, unintelligible word attempt, sign, or recognizable word was transcribed regardless of pronunciation accuracy. To assure accuracy of the language sample transcripts, a word-by-word, inter- and intrajudge transcription reliability was calculated for 20% of each transcript. Reliability ranged from 88 to 94% agreement, indicating good agreement. Vocabulary was analyzed using the number of different words produced in the language samples at 12, 18, 24, and 30 months of age. The number of different vocabulary words used in natural conversation is a highly sensitive measure of early language development.

Parent Report of Language Development. To validate the language sample findings, a parent report of language development was obtained using the Communicative Development Inventory (CDI) [Fenson et al., 1993] at 12, 18, 24, and 30 months of age. The CDI is a normed parent-report measure that assesses a child's vocabulary size, length of their three longest sentences, and use of word endings. The validity of the CDI has been determined for typically developing children, children with delayed speech and language development, and children having CLP and ICP [Dale, 1991; Scherer and D'Antonio, 1995].

Speech Sound Production. In English, consonants are the sounds that carry the most information regarding word meaning. Children who are normally developing acquire sounds in a well-ordered manner. Sound development has been documented by describing stages of babbling that continue into meaningful word use [Stoel-Gammon, 1989]. Children progress from use of vowel's and consonant-like sounds (for example, w, y) to use of true consonants (such as p, t, k, m, s, f). The number of sounds children use begins to increase before words begin (between 6 and 12 months of age) but expands rapidly with early word use. By the time normal developing children are 2 years old they are using 12 to 14 different consonant sounds in their speech [Stoel-Gammon, 1989]. The use of consonant and vowel combinations, referred to as syllable structure, advances in complexity from similar consonants and vowels (e.g., mama, dada) to varied consonant and vowel patterns (e.g., banky, doggy). This developmental pattern is altered often when there is a structural abnormality such as clefting. Children with clefts may use fewer or different sounds [O'Gara and Logemann, 1988].

For this study, speech was assessed by transcribing the vocalizations and word attempts of the children from the video-taped language samples that included a

naming activity with a set of toys controlled for sound features. Sounds were considered present in the child's repertoire if they occurred at least three times [Stoel-Gammon, 1985]. Each word attempt or vocalization was transcribed following the rules developed by Paul and Jennings [1992]. Consonant inventories were derived by counting all consonants that met the above criteria.

Particular emphasis was placed on the accurate transcription of compensatory articulation errors, particularly glottal stops. It is possible that word attempts that are comprised of a glottal stop and a vowel may be perceived as an isolated vowel production. Special training was given to the transcribers to increase their sensitivity to the presence of glottal stop substitutions. In instances where an isolated vowel was transcribed, the videotapes were reviewed to check whether a glottal stop or other compensatory articulation may have preceded the vowel.

Percent Consonant Correct-Revised (PCC-R) [Shriberg et al., 1997] was used as a measure of consonant accuracy in connected speech at the 30-month evaluation. 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 in the sample. Only consonant substitutions and omissions were considered as errors, whereas consonant distortions caused by nasal emission or dental deviations are ignored in this analysis. This metric of articulation competence had 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 severity of speech impairment (normal, mild, moderate, or severe impairment).

Velopharyngeal Function. One component of communication that is frequently affected in children with palatal clefts or VCFS is velopharyngeal (VP) function. Hypernasality and nasal emission are the characteristics most commonly associated with inadequate VP function. It is difficult to obtain definitive measures of VP function during early speech development; however, preliminary perceptual judgments can be made from early speech sound production. For purposes of this study, perceptual ratings of VP function were made from composite ratings of hypernasality and nasal emission during oral consonant attempts. Ratings were initiated when the child had at least three different words and were then made at each successive evaluation from word attempts on the language sample. VP symptoms were rated on a 1 to 7 equal appearing interval scale with 1 representing the presence of mild VP symptoms and 7 representing severe symptoms.

Data Analysis

Group values at each time point were summarized by the mean (Figures) and standard deviation (Table III). Statistical analysis was performed with analysis of variance (ANOVA) to assess the four groups, time periods, and the interaction of the group and time factors. Since group main effects and all interaction effects were significant for each variable ($p < 0.05$), the least significant difference procedure was used to compare group means at each time point.

TABLE III. Mean Standard Score and Statistical Comparison of the Children With VCFS, ICP, CLP, and Noncleft Children on the Motor and Mental Scales of the Bayley Scales of Infant Development*

	Age in months		
	6	18	30
		Motor scale	
VCFS	91.0 ± 11.3	84.3 ± 18.0	82.5 ± 20.1
ICP	78.6 ± 13.0	83.4 ± 17.0	84.2 ± 20.9
CLP	98.1 ± 9.1	100.1 ± 14.7	105.9 ± 8.1
Normal	106.4 ± 11.7	108.8 ± 7.3	110.0 ± 7.6
VCFS vs. ICP	NS	NS	NS
VCFS vs. CLP	NS	NS	$p = 0.01$
VCFS vs. normal	NS	$p = 0.01$	$p = 0.01$
		Mental Scale	
VCFS	89.2 ± 18.1	80.0 ± 8.5	67.2 ± 13.7
ICP	86.0 ± 11.3	77.6 ± 14.1	77.0 ± 20.7
CLP	99.6 ± 8.3	99.1 ± 13.5	102.1 ± 8.1
Normal	102.5 ± 7.5	106.8 ± 10.3	108.2 ± 11.5
VCFS vs. ICP	NS	NS	NS
VCFS vs. CLP	NS	$p = 0.05$	$p = 0.01$
VCFS vs. normal	NS	$p = 0.01$	$p = 0.01$

*VCFS = children with velocardiofacial syndrome; ICP = children with isolated cleft palate; CLP = children with cleft lip and palate; normal = children without clefts; NS = not significant. Summary statistics are mean ± standard deviation.

RESULTS

Developmental Measures

Motor Development. Performance of the children with VCFS is compared with data from the three comparison groups. The upper panel of Table III shows the mean performance and results of statistical comparisons for the standard scores of the motor subscale of the BSID. The individual performance of the children with VCFS showed that three children were in the normal range and one child fell into the delayed range. The one child who appeared to show delays also had behavioral issues (e.g., noncompliance to structured tasks, short attention to activities) that were believed to affect testing performance. The motor scores were significantly lower than the noncleft children ($p < 0.05$) and lower than the children with CLP by 30 months of age ($p < 0.05$). However, scores for the children with VCFS were not statistically different from the performance of the children with ICP at any age.

Mental Development. In the lower panel of Table III the mean performance and statistical comparisons of the children with VCFS are shown. When viewing the language and nonlanguage items combined, the scores of the children with VCFS were statistically lower than those of the noncleft children ($p < 0.05$) and children with CLP ($p < 0.05$) but not from those of the children with ICP ($p = 0.37$). Inspection of the mental scale scores for the children with VCFS showed what might be interpreted as a decline in scores from 18 months through the 30-month assessment. Because it has been determined that the children with VCFS have lan-

guage impairments, this finding may reflect the increasing contribution of language items in tests during this developmental period. Therefore, the language and nonlanguage items were examined separately. The language/nonlanguage performance for the children with VCFS at 30 months is presented in Table IV. The data indicate that all the children with VCFS showed poorer performance on the items that required language than items that did not require language. Of the 106 cognitive (nonlanguage) items, the children with VCFS passed 106, 103, 100, and 81 items, respectively, indicating that the full mental scale score is not reflective of nonlanguage cognitive performance.

General Measures of Language Development

The children with VCFS showed receptive-expressive language impairments that were apparent from the onset of language. Further, the course of this early language impairment persisted and widened when compared with the development of children with CLP and ICP. Figure 1 shows the receptive language age scores obtained for the children with VCFS, ICP, CLP, and children without clefts from 6 to 30 months of age. ANOVA and pairwise comparison of group means indicated that performance of the children with VCFS was poorer than the children without clefts from 12–30 months (p <0.05). Further, when compared with the children with CLP and ICP, the children with VCFS fell behind those groups by 24 months of age (p <0.05). These data indicate that receptive language was impaired from the onset of language and the delay widened through 30 months of age.

Figure 2 shows the expressive language performance for the children with VCFS, normally developing children, and children with CLP and ICP. The performance of the children with VCFS was statistically different from the children without clefts from 12 months onward (p <0.05). By 24 months of age, the children with VCFS were different from the children with CLP (p <0.05) and by 30 months, their performance fell below that of the children with ICP (p <0.05).

Language Sample

The number of different words, a measure of vocabulary use, is presented in Figure 3. The children with VCFS show severe restrictions in their vocabulary use when compared with all three comparison groups. Statistical comparisons revealed that the vocabulary use of the children with VCFS was significantly poorer than that of the children without clefts (p <0.05) and children with CLP by 24 months (p <0.05) and poorer than that of the children with ICP by 30 months of age (p <0.05).

It should be noted that all word attempts or signs were credited in the coding. This is an important methodological feature of this study in that every communicative attempt, through any modality, was given credit in the analysis.

TABLE IV. Language and Nonlanguage Item Analysis From the Bayley Scales of Infant Development-2 (BSID-2) for the Children With VCFS

Subject	BSID-2 Mental	BSID-2 Motor	Number of language items passed	Number of cognitive items passed
1	68	80	15/49	103/106
2	<50	55	13/49	81/106
3	77	83	17/49	106/106
4	63	80	15/49	100/106

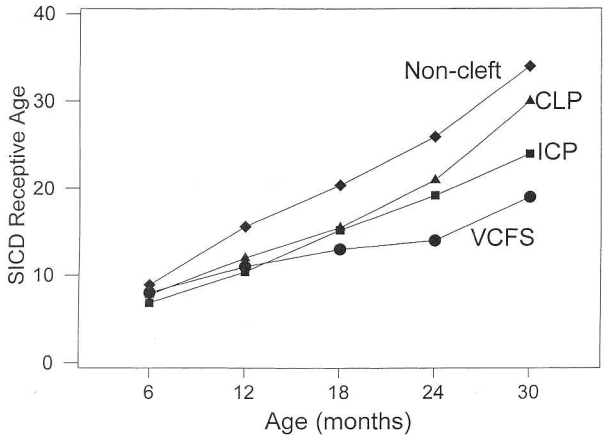

Fig. 1. The receptive age scores from the *Sequenced Inventory of Communicative Development-R* are presented at 6, 12, 18, 24, and 30 months of age for the noncleft children (diamonds), children with cleft lip and palate (triangles), children with isolated cleft palate (squares), and children with VCFS (circles).

Parent Report Measures of Language Development

The language sample provides one view of the child's language use but does not represent the total vocabulary that the child may know. A parent report measure of vocabulary size was collected using a standardized form, the MacArthur Communicative Development Inventory. Figure 4 shows the number of different words indicated by the parent from 12 to 30 months of age. The vocabulary size profile for the children with VCFS was flat, indicating that vocabulary size was extremely

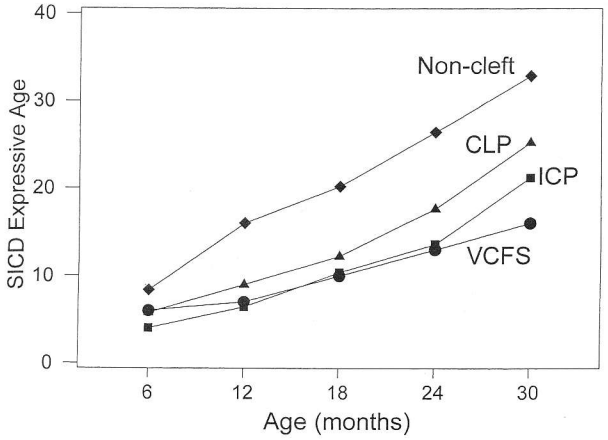

Fig. 2. The expressive age scores from the *Sequenced Inventory of Communicative Development-R* are presented at 6, 12, 18, 24, and 30 months of age for the noncleft children (diamonds), children with cleft lip and palate (triangles), children with isolated cleft palate (squares), and children with VCFS (circles).

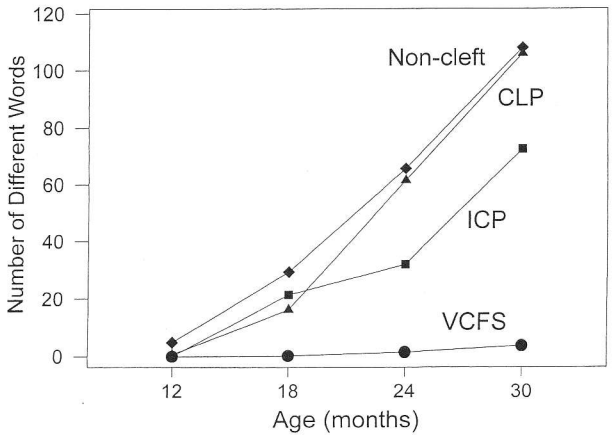

Fig. 3. The number of different words used in the language sample as derived from the *Systematic Analysis of Language Transcripts* are presented at 6 through 30 months of age for the noncleft children (diamonds), children with cleft lip and palate (triangles), children with isolated cleft palate (squares), and children with VCFS (circles).

small and did not show the rapid growth pattern expected at this age. Statistical comparison of the profiles showed that the vocabulary size of the children with VCFS was poorer than the noncleft children, beginning by age 18 months (p <0.05); differed from the children with CLP by 24 months (p <0.05); and differed from the children with ICP by 30 months of age (p <0.05). These findings are similar to those obtained from the language sample. It is important to note the severely restricted communicative performance of the children with VCFS. These children were without even the most functional vocabulary during a critical period for vocabulary growth.

Speech Sound Production

This study measured the number of consonant types that approximated the adult form of the words and any consonant vocalization produced by the children. The number of consonant types used was examined in the language sample and is displayed in Figure 5 and Table V. The children with VCFS showed severely restricted consonant inventories when compared with the three comparison groups. Statistical comparisons indicated that the number of consonant types used in words or word attempts by the children with VCFS was lower than that of the noncleft children and children with CLP by 18 months (p <0.05) and differed from the children with ICP by 24 months of age (p <0.05). In addition to severe limitations in consonant inventory, children with VCFS had significantly lower PCC-R ratings (p <0.05) than the children with CLP, ICP and those in the noncleft group. PCC-R ratings indicated that articulation accuracy for the children with VCFS was severely delayed for their age. Further, the children with VCFS used glottal stop substitutions more often than children in the CLP or ICP groups (p <0.05).

Velopharyngeal Function

Results of the perceptual ratings of VP function are presented in Table V. None of the children in the normally developing group demonstrated VP symptoms. Approximately 50% of the children in the CLP and ICP groups demonstrated some mild/moderate VP symptoms, and all of the children in the VCFS group were perceived as having some hypernasality and/or nasal emission. Another factor that has been suggested as being related to the presence, type, or severity of speech errors in children with cleft palate is the age at palate repair. The results provided in Table V demonstrate there is no apparent relationship between the age at time of palate repair and the number of consonant types, PCC-R rating, or percentage of glottal stop substitutions for the children in this sample.

The relationship between VP function and the types and pattern of speech errors was also assessed. Table V shows the rating of VP symptoms, number of consonant types, PCC-R rating, and percentage of glottal stop substitutions for subjects in all groups. Inspection of

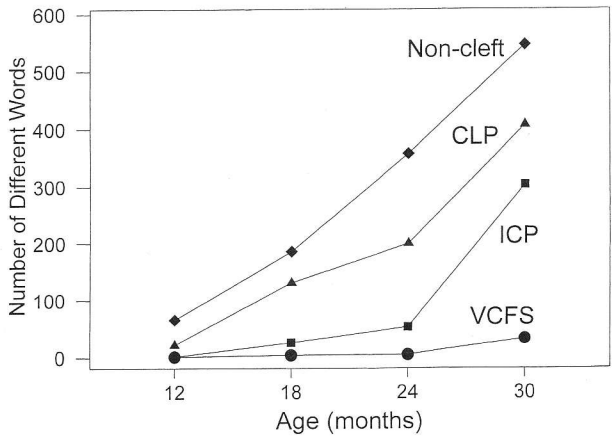

Fig. 4. The number of different words used from the parent report measure, the *MacArthur Communicative Development Inventory*, are presented at 6 through 30 months of age for the noncleft children (diamonds), children with cleft lip and palate (triangles), children with isolated cleft palate (squares), and children with VCFS (circles).

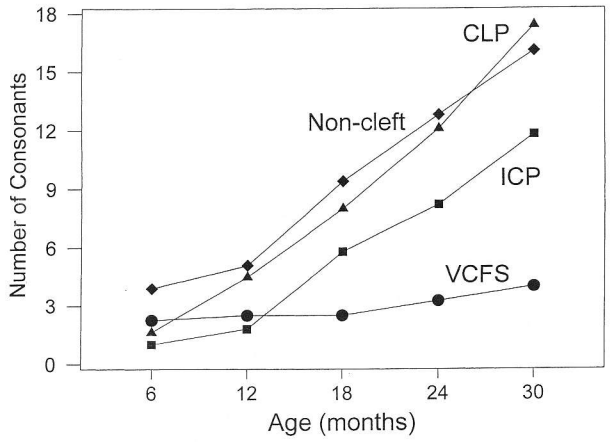

Fig. 5. The number of consonants used as derived from the *Systematic Analysis of Language Transcripts* are presented at 6 through 30 months of age for the noncleft children (diamonds), children with cleft lip and palate (triangles), children with isolated cleft palate (squares), and children with VCFS (circles).

TABLE V. Age at Palate Repair, Velopharyngeal Symptoms, and Articulation Variables for Children in the Four Groups at 30 Months of Age

Subject	Age at palate repair	VP symptoms	Consonant types	PCC-R rating	%Glottals
Normal					
1	NA	None	21	76	0
2	NA	None	16	71	0
3	NA	None	15	68	5
4	NA	None	18	91	0
5	NA	None	15	88	0
6	NA	None	15	72	0
7	NA	None	18	92	0
8	NA	None	15	64	10
Mean			16.6	77.7	7.5**
CLP					
1	15 M	None	16	73	4
2	12 M	Moderate	19	46	25
3	12 M	None	15	63	7
4	15 M	Moderate	13	58	12
5	15 M	None	18	70	9
6	12 M	Moderate	10	60	23
7	12 M	None	22	69	0
8	12 M	Mild	16	46	15
Mean			16.1	60.6	13.6
ICP					
1	12 M	None	21	71	8
2	12 M	Moderate	7	30	12
3	12 M	Mild	11	50*	12
4	12 M	None	13	57	3
5	15 M	Mild	4	59*	15
6	15 M	Moderate	5	72*	20
7	12 M	Mild	15	67	5
Mean			10.8	58	10.7
VCFS					
1	—	Mild–Moderate	3	46*	23
2	12 M	Mild	2	41*	25
3	—	Mild	5	40*	18
4	13 M	Mild	5	47*	15
Mean			3.7	43.5	20.3
Comparison of means					
NC vs. CLP		NS	$p < 0.05$		$p < 0.05$
NC vs. ICP		$p < 0.05$	$p < 0.05$		$p < 0.05$
NC vs. VCFS		$p < 0.05$	$p < 0.05$		$p < 0.05$
CLP vs. ICP		$p < 0.05$	NS		NS
CLP vs. VCFS		$p < 0.05$	$p < 0.05$		$p < 0.05$
ICP vs. VCFS		$p < 0.05$	$p < 0.05$		$p < 0.05$

NA = not applicable; M = Months of age; * = PCC-R calculated on 10 words or fewer; ** = mean % glottals calculated for only those subjects who used glottal sounds; NS = not significantly different; CLP = cleft lip and palate; ICP = isolated cleft palate; VCFS = velocardiofacial syndrome.

the data indicates there is no apparent relationship between the presence or severity of VP symptoms and the number of consonant types or PCC-R rating. On the other hand, there does appear to be a relationship between the presence of VP symptoms and the percentage of glottal stop consonants used. That is, the children with CLP and ICP with no VP symptoms had a lower frequency of glottal stop articulation than the children with mild or moderate VP symptoms. However, among the children who did demonstrate VP symptoms, the percentage of glottal stop consonants used did not appear to be related to the severity of VP symptoms. Furthermore, the VCFS group showed a significantly higher frequency of glottal stop use than children with CLP ($p < 0.05$) or ICP ($p < 0.05$) regardless of the severity of VP symptoms.

DISCUSSION

Speech and Language Characteristics of Young Children with VCFS

The purpose of this study was to provide a longitudinal description of several critical features of early speech and language development for a small sample of children with VCFS from 6 to 30 months of age. The profile observed for the children with VCFS was compared with patterns of development for typically developing children, children with cleft lip and palate, and children with isolated cleft palate. Inspection of the data reveals a hierarchy of performance, with the noncleft children performing the best, followed by the children with CLP, then children with ICP, with the children with VCFS consistently showing poorest performance on all measures of speech–language develop-

ment. The present study also suggests that the developmental profile of the speech and language impairment observed in children with VCFS appears to differ from that of typically developing children as well as the two groups of children with clefts.

The data indicate that the children with VCFS demonstrated severe receptive–expressive language impairments from the onset of language, and these impairments increased in severity from 12 to 30 months of age. Furthermore, for the children in this study, both early vocabulary and speech sound acquisition were severely impaired to the extent that the children were essentially nonoral through 30 months of age. There is a marked discrepancy between receptive language performance and expressive language and speech production. Also, the expressive impairments were not consistent with the performance on the mental scale of BSID.

Comparison With Data From Cross-Sectional Studies

Early reports describing VCFS identified speech–language and learning impairments as prominent features of the syndrome [Golding-Kushner et al., 1985; Shprintzen et al., 1978]. However, since these first publications, there have been few reports that describe the nature and extent of such impairments. The present study is the first longitudinal assessment of speech and language development in children with VCFS. Additionally, most previous studies have focused on older children, whereas the present study described communication development in children from birth to age 3. Because of the early age of this population and differences in methodology, the data from this study provide important information that identifies features that may be the precursors to the speech, language, and learning characteristics described in older children with VCFS.

For example, the cross-sectional studies in the literature are in conflict regarding whether language impairments in young children with VCFS improve during the preschool years [Solot et al., 1998] or persist into school age [Nayak and Sells, 1998]. Solot et al. discussed the presence of a receptive–expressive language deficit that appeared to be improving for her group of 40 preschool children with VCFS. However, Nayak and Sells [1998] found that 89% of their school-aged group showed persistent language impairments. Data from the present study suggest that deficits in language increase in severity from birth to 3 and become distinct from typically developing children and children with clefts by 12 to 18 months of age. These data show that speech and language impairments in children with VCFS occur earlier than has been reported previously. Also, these impairments become readily identifiable when methods are used that are sensitive to the profile of language impairment seen in these children. For example, data from this study indicate that the most severe component of language impairment for young children with VCFS was evident in language use observed within natural communicative contexts. Previous studies of language development have relied on standardized tests of language performance. Generally, the severity of impairment seen in the present population is not readily apparent when general tests of speech–language development are used, as they do not take into consideration the child's ability to use language in real communicative settings. Therefore, impairments such as those observed in this study and the substantial differences from typically developing children may not have been apparent in previous studies.

Previous reports have speculated regarding the relationship between speech and language and cognitive function in children with VCFS. [Golding-Kushner et al., 1985; Swillen et al., 1997; Solot et al., 1998]. However, there is no consensus regarding the nature of this relationship. Data from the present study show that expressive language impairments were not consistent with the performance on the mental scale of BSID. While the children showed increasing deficits in expressive language from 6 through 30 months of age, when the language items from the BSID were separated from the nonlanguage items, performance on the nonlanguage items was always superior to performance on language items. This finding suggests that these children demonstrated language impairments that exceeded cognitive deficits.

If cognitive delay is not solely responsible for the early speech and language impairments seen in children with VCFS, then what are the possible hypotheses regarding the bases of these early delays? One possible explanation may be that these characteristics are similar to children with other genetic syndromes. Characteristic profiles of developmental and speech–language impairments in children with a number of syndromes have been reported, as in Down syndrome [Miller, 1988] or Williams syndrome [Karmiloff-Smith et al., 1997]. However, the characteristics described in this study for children with VCFS do not match documented developmental profiles for other syndromes. For example, children with Down syndrome or Williams syndrome typically show a more significant cognitive impairment than observed in children with VCFS. One explanation may be that at least some children with VCFS have a unique developmental profile consisting of mild cognitive deficits, receptive–expressive language impairment, with expressive language and speech sound production severely delayed. The unique feature of this profile is the severity of early expressive language and speech sound production impairments relative to other developmental parameters.

Another characteristic of early speech and language development observed in this study was the severity of the expressive language and speech impairment in the children with VCFS. For the children in this study, both early vocabulary and speech sound acquisition were severely impaired to the extent that the children with VCFS were essentially nonoral through 30 months of age. Solot et al. [1998] also discussed the disparity between expressive and receptive language performance in their cross-sectional study of preschool children. Some of the children described in the Solot study were nonoral. However, the children were observed at only one point in their development, so it is unclear how long they remained at that level and what the course of development had been prior to and fol-

lowing that single observation. The present data would suggest that these severe expressive impairments occur from the onset of language and can be identified as early as 18 months of age.

Another factor that has been identified as a precipitating factor in the speech–language patterns observed in children with VCFS is the high occurrence of VP dysfunction. Solot et al. [1998] suggested that palatal function was not the sole factor responsible for the profile of speech and language performance of the children they observed with VCFS. The present study provides data concerning the relationship between VP function and speech production. An important feature of the present study is the use of the comparison groups of children with cleft lip and palate and isolated cleft palate. Data from this study clearly demonstrate that the children with VCFS not only differed from typically developing children but they also showed more impairments in speech and language development than the two comparison groups of children with palatal clefting. Furthermore, the present data do not support a simple causal relationship between the severe speech production abnormalities observed for the children with VCFS and the presence of VP dysfunction. The VCFS group demonstrated significantly greater speech production deficits than children in the two cleft groups who also experienced VP symptoms. The present data clearly indicate that the relationship between VP function and speech sound errors is not as simple and straightforward as has been suggested previously.

Directions for Future Research

This study has provided detailed information regarding the early communicative characteristics of a small sample of children with VCFS. The present findings interpreted in conjunction with the existing literature point to a distinctive profile of speech and language impairment for children with VCFS. However, several important questions remain unanswered. For example, it is not known whether there are subgroups within this profile that may account for some of the differences described in the literature and anecdotally. Also, it is not clear what proportion of children with VCFS have these profiles of speech and language impairment or how they change over time. Golding-Kushner [1985] and McDonald-McGinn et al. [1997] documented the presence of learning disabilities in a school-aged population of children with VCFS. In other populations of children, early language impairment has been identified as a precursor to later learning disabilities. Therefore, future investigations should consider whether the learning disabilities described in children with VCFS have their origins in early language and speech impairments.

Another area of controversy that warrants further study is the course of the early speech production deficits. Do they improve with age and/or with treatment? Solot et al. [1998] suggest that all but two of their study groups were speaking by school age, suggesting that the speech production deficits may improve. Also, how do the early speech production deficits influence later language and learning deficits observed in these children at school age? Do limitations in speech production reduce the possible options for expressing different words, thereby contributing to a delay? Or do early language impairments limit the need to attempt a variety of sounds? What is the relationship between VP dysfunction and the type and severity of speech sound errors in children with VCFS? If there is not a strong relationship between VP symptoms and speech errors, what accounts for the high occurrence of compensatory articulation patterns, particularly the high percentage of glottal stop substitutions?

Another area that should be addressed is the relationship between the severe early language and speech impairment and the behavioral or psychosocial problems apparent for many children with VCFS. What are the relationships between inheritance, environmental factors, and speech and language performance? These questions require future longitudinal studies with larger numbers of subjects. Answers to these questions would provide valuable information for developmental monitoring and early intervention strategies for children with VCFS.

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