Published in final edited form as:
J Speech Lang Hear Res. 2009 August ; 52(4): 1048–1061. doi:10.1044/1092-4388(2009/08-0001).

Phonological Accuracy and Intelligibility in Connected Speech of Boys with Fragile X Syndrome or Down Syndrome

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Abstract

Purpose—We compared the phonological accuracy and speech intelligibility of boys with fragile X syndrome with autism spectrum disorder (FXS-ASD), fragile X syndrome only (FXS-O), Down Syndrome (DS), and typically developing (TD) boys.

Method—Participants were 32 boys with FXS-O (3 to 14 years), 31 with FXS-ASD (5 to 15 years), 34 with DS (4 to16 years), and 45 TD boys of similar nonverbal mental age. We used connected speech samples to compute measures of phonological accuracy, phonological process occurrence, and intelligibility.

Results—The boys with FXS, regardless of autism status, did not differ from TD boys on phonological accuracy and phonological process occurrence but produced fewer intelligible words than TD boys. The boys with DS scored lower on measures of phonological accuracy and occurrence of phonological processes than all other groups and used fewer intelligible words than TD boys. The boys with FXS and the boys with DS did not differ on measures of intelligibility.

Conclusion—Boys with FXS, regardless of autism status, exhibit phonological characteristics similar to those of younger TD children but are less intelligible in connected speech. The boys with DS show greater delays in all phonological measures than the boys with FXS and TD boys.

Keywords

Fragile X Syndrome; Down Syndrome; Autism; Phonology

Introduction

Fragile X syndrome (FXS) and Down syndrome (DS) are the two most common genetic causes of intellectual disability (Dykens, Hodapp, & Finucane, 2000; Hagerman & Hagerman, 2002). In addition to a wide range of cognitive deficits, poor speech intelligibility has been reported as one of the most common communication characteristics of children with these syndromes (Abbeduto & Hagerman, 1997; Dodd & Thompson, 2001; Miller & Leddy, 1999; Stoel-Gammon, 1997, 2001). Although descriptions of the cognitive phenotypes of these

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children have been frequently reported in recent years (Churchill et al., 2002; Dykens et al., 2000; Hagerman & Hagerman, 2002; Kau, Meyer, & Kaufmann, 2002; McElwee & Bernard, 2002; Prasher & Cunningham, 2001; Pueschel, 1994; Roizen, 1997), specific details about their communicative phenotype have not been as well documented. Poor speech intelligibility has been reported for both groups, with most studies of articulation and phonology based on either single word production accuracy (Dodd & Thompson, 2001; Hanson, Jackson, & Hagerman, 1986; Kumin, 2001; Paul et al. 1987; Prouty et al., 1988; Smith & Stoel-Gammon, 1983; Stoel-Gammon, 2001) or in the case of FXS, case reports (Madison, George, & Moeschler, 1986; Palmer, Gordon, Coston, & Stevenson, 1988; Paul, Cohen, Breg, Watson, & Herman, 1984). There are no studies regarding the connected speech characteristics of children with FXS, and most studies on this topic in DS are based solely on parental report (Berglund, Eriksson, & Johansson, 2001; Kumin, 1994).

FXS is an X-linked genetic condition and the most common inherited cause of intellectual disability, with one in every 4,000 individuals being affected (Crawford, Acuna, & Sherman, 2001; Turner, Webb, Wake, & Robinson, 1996). Although FXS can affect both genders, the cognitive, communicative and behavioral phenotypes differ greatly for males and females (Abbeduto & Hagerman, 1997). Males are typically more severely affected than females, exhibiting more marked delays in several areas of development (Abbeduto & Hagerman, 1997; Abbeduto, Murphy, et al., 2003; Hagerman & Hagerman, 2002). Intellectual disability, ranging from mild to profound, and deficits in language and phonological skills are common among males with FXS, whereas females are usually less affected in their cognitive and communication skills (Abbeduto, Brady, & Kover, 2007; Abbeduto & Hagerman, 1997; Abbeduto, Murphy, et al., 2003; Palmer et al., 1988; Rice, Warren, & Betz, 2005; Spinelli, Rocha, Giacheti, & Richieri-Costa, 1995). There is also a higher prevalence of autism in males with FXS (Clifford et al., 2006), which often co-occurs with other cognitive and communicative disorders. Due to these differences in the phenotypes of males versus females with FXS, only males participated in the present study.

Previous research has described the speech intelligibility and speech sound errors characteristic of boys with FXS at the single word level. Hanson and colleagues (1986) reported that ten boys with FXS (ages 3 to 9 years) exhibited sound substitutions, omissions, and distortions on a single word articulation test. Prouty and colleagues (1988) also reported that all but one of the fifteen males (ages 3 to 23 years) exhibited common developmental errors of substitution and omission. Madison and colleagues (1986) reported on the speech production accuracy of five male family members with FXS (ages 4 to 34). Although some substitution and omission errors were noted among these family members, articulation scores were in the normal range, and the males were reported to be intelligible when speaking in one or two word utterances. Paul and colleagues (1984) reported that three boys with FXS (ages 10 to 13) exhibited developmental phonological processes such as liquid simplification and final consonant deletion but that their speech was intelligible at the single word level. In a more recent study, Roberts and colleagues (Roberts et al., 2005), compared single word phonological accuracy and phonological process occurrence in 50 boys with FXS (ages 3 to 14 years), 32 boys with DS (ages 4 to 13 years), and 33 younger, mental-age-matched TD boys (ages 2 to 6 years) and found that the boys with FXS, although delayed in their speech development, did not differ from the younger TD boys in their percentage of correct consonants, phonological processes, and whole word proximity scores. These studies indicate that the phonological patterns of males with FXS at the single word level are similar to the sound patterns used by younger children at the same developmental level. Given the present lack of research regarding phonology in the connected speech of children with FXS, the purpose of the current study is to determine whether the speech production characteristics in connected speech are different from those found at the single word level.

In the present study, boys with FXS were divided into two groups: those with and without a concurrent diagnosis of ASD. Some research suggests that speech production is not adversely affected in children with autism who use verbal language when compared to other children of similar mental or linguistic age (Bartolucci & Pierce, 1977; McLeery, Tully, Sleve, & Schreibman, 2006). For example, McCleery and colleagues found that 14 children with ASD (ages 2 to 6 years) exhibited a similar pattern of consonant production as TD children (ages 13 to 14 months) at similar linguistic ages. However, there are other reports that children with autism exhibit more motor speech difficulties and speech production delays when compared to TD children of the same chronological age (Adams, 1998; Shriberg et al., 2001). Given that reduced speech intelligibility has been reported in children with FXS, it is possible that children with both FXS and ASD might exhibit more serious speech production deficits than children with FXS alone. Given a high reported prevalence of autism among males with FXS (15-25%; Bailey et al., 1998; Dykens & Volkmer, 1997; Hagerman, 2002), one of our study objectives was to determine whether or not co-morbidity of ASD affects the speech production of children with FXS, therefore our participants with FXS were divided according to autism status.

The most common genetic cause of intellectual disability, Down syndrome has a prevalence of 13.65 per 10,000 live births (Carothers, Hecht, & Hook, 1999; Centers for Disease Control and Prevention, 2006). There is a wide range of cognitive impairment in this population, although up to 80% of individuals with DS display moderate to severe intellectual disability (Prasher & Cunningham, 2001; Pueschel, 1994; Roizen, 1997). In addition to having similar levels of intellectual disability, boys with DS and boys with FXS are both reported to have varying degrees of deficits in speech intelligibility, articulation and phonological skills, and expressive language. There are many reports of phonological deficits and impaired speech intelligibility in children with DS (Bleile & Schwarz, 1984; Dodd & Thompson, 2001; Kumin, 1994; Smith & Stoel-Gammon, 1983; Stoel-Gammon, 1980, 1997, 2001). In a study of the production of stops, Smith and Stoel-Gammon (1983) found that five children with DS (ages 3 to 6) displayed common developmental phonological processes such as cluster simplification, final stop devoicing, and final consonant deletion but suppressed their usage at a slower rate than TD children. Other researchers also found that children with DS displayed common developmental phonological processes such as deletion of final consonants, cluster reduction, stopping, and liquid simplification (Bleile & Schwarz, 1984; Stoel-Gammon, 1980). Kumin and colleagues, who studied the emergence of phonemes in 60 children with DS (ages 9 months to 9 years) found that although they emerged later, the phonemes emerged in a similar order to that of TD children with few exceptions (Kumin, Councill, & Goodman, 1994). These studies suggest that the majority of children with DS develop speech at a slower rate but follow the same patterns of development as TD children. One study, however, found that ten six- to fourteen-year-old children with DS used phonological processes more often and displayed phonological processes that were not displayed by TD children (Dodd, 1976). Another study reported that 15 children with DS (ages 5 to 15) exhibited more inconsistency in phoneme production than 15 phonologically delayed but otherwise TD children; this finding lends support to evidence of a phonological disorder rather than a delay (Dodd & Thompson, 2001). Likewise, Roberts and colleagues (2005) found that boys with DS were more delayed in their mastery of consonant phonemes, phonological process occurrence, and whole word proximity scores when compared to mental-age-matched younger TD boys. The nature of the phonological deficits underlying impaired intelligibility in children with DS needs further study, especially in regards to the connected speech characteristics of children with DS. Including participants with DS in this study will not only further our knowledge about their phonological accuracy in connected speech, but will also help to determine whether the deficits in intelligibility, articulation and phonological accuracy in children with FXS and DS are due to the presence of intellectual disability in general, or whether there are differences in these skills that are idiosyncratic to each group's individual phenotypes. Finally, it will help determine whether there are different relationships between phonological accuracy in single

words versus connected speech for each group, and how these skills relate to speech intelligibility.

Connected speech is considered by some researchers to be a more valid context than single words for evaluating phonological delay and speech intelligibility (Hoffman & Norris, 2002; Flipsen, 2006). Regardless of a child's particular diagnosis, speech production in connected speech may differ from speech production on single word, standardized tests. For example, Iacono (1998) compared the phonological skills of children with DS in single-word versus connected speech samples. Although there was no significant difference in the accuracy of consonants across conditions, connected speech samples yielded fewer productions of later developing phonemes, total words, and word tokens than single word articulation tests (Iacono, 1998). The author concluded that some children with DS might avoid phonemes in connected speech that they had yet to master. Similarly, Morrison and Shriberg reported that speech production analyses based on connected speech samples of 61 speech-delayed children (ages 4 to 6 years) were significantly different from that of single word articulation tests, concluding that connected speech may be a more sensitive context for assessing less well-established or later developing phonemes and motor speech skills than single-word articulation tests (Morrison & Shriberg, 1992). Two other studies found mixed results, reporting that in speech-impaired children of typical intelligence, phonological processes occurred significantly more often in connected speech than in single word productions, but that overall phonological accuracy was more similar than different across the contexts (Andrews & Fey, 1986; McLeod, Hand, Rosenthal, & Hayes,1994).

Given that Roberts and colleagues (2005) found no significant difference between the phonological accuracy of single word productions of boys with FXS as compared to that of their TD peers and the frequent reports of reduced intelligibility in boys with FXS (Abbeduto & Hagerman, 1997; Hanson et al., 1986, Prouty et al., 1988), the present study is a follow-up to Roberts et al., 2005 to determine whether group differences not found in single words might be found in connected speech. We examined speech production accuracy in spontaneous, connected speech in order to determine whether the phonological accuracy, occurrence of phonological processes, and intelligibility of boys with FXS or DS differed from those of developmentally similar TD boys and whether autism status was a significant factor in the connected speech of boys with FXS. Given the many reports of reduced intelligibility in children with FXS and DS, we expected the participants in these populations to score lower than the TD boys in phonological accuracy, phonological process occurrence, and intelligibility in connected speech. Further, given the reported speech production deficits in children with ASD, we expected the boys with FXS-ASD to score lower than the boys with FXS-O on these outcomes.

Methods

Study Population

Table 1 describes study participants, which included four groups of boys at similar nonverbal mental ages. The groups were 32 boys with a diagnosis of FXS only (FXS-O), 31 boys with FXS with a co-occurring diagnosis of ASD (FXS-ASD), 34 boys with DS (DS), and 45 typically developing (TD) boys. To be eligible for study participation, all boys with FXS (with and without ASD) and boys with DS were between 3 and 14 years of age with an expressive vocabulary of at least 40 words, combining at least two words (MLU greater than 1.1), and passed a pure tone hearing screening at 25 dB HL in the better ear at 500, 1,000, 2,000, and 4,000 Hz. All participants used verbal communication (rather than sign) and spoke English as the primary language at home. Boys with DS were excluded from participation if they had a previous diagnosis of ASD or if they scored in the autism or autism spectrum range on the *Autism Diagnostic Observation Schedule - General* (ADOS; Lord, Rutter, DiLavore, & Risi,

2002). Exclusionary criteria for the TD group were a diagnosis of intellectual disability, developmental disability, ASD, nondevelopmental speech or language deficits; if they scored less than 1.5 SD below the mean on tests of nonverbal intelligence, speech, or language; or if they were enrolled in speech-language services. Study participant recruitment procedures were described in detail by Roberts and colleagues (2005).

Study procedures were approved annually by the School of Medicine Institutional Review Board at the University of North Carolina at Chapel Hill. Parental informed consent was obtained at or before the time of the first study assessment.

Table 1 describes the four groups of study participants:

Fragile X syndrome only, without autism spectrum disorder (FXS-O)—This group included 32 boys with FXS without a diagnosis of ASD ranging in chronological age from 3.2 years to 14.5 years (M = 10.9 years), with a mean nonverbal cognitive age equivalent of 5.3 years on the *Leiter-R*. All boys with FXS were required to have a diagnosis of full mutation FXS, confirmed by DNA analyses, in order to participate in the study. The boys with FXS were included in this group if they did not score within the autism or autism spectrum range on the ADOS administered at the first assessment time point. Eighty-four percent of the participants were Caucasian, 13% were African American, and 3% reported another ethnicity. Thirty-eight percent of the mothers of the participants in this group had a terminal education level of a high school degree, and 62% had some college or a college degree.

Fragile X syndrome with autism spectrum disorder (FXS-ASD)—This group included 31 boys with FXS with a diagnosis of ASD ranging in chronological age from 5.0 years to 15.5 years (M = 10.12 years), with a mean nonverbal cognitive age equivalent of 5.1 years on the *Leiter-R*. All boys with FXS were required to have a diagnosis of full mutation FXS, confirmed by DNA analyses, in order to participate in the study. The boys with FXS were included in this group if they scored within the autism or autism spectrum range on the ADOS administered at the first assessment time point. Ninety percent of the boys were Caucasian and 10% were African American. Nineteen percent of the mothers of the participants in this group had a terminal education level of a high school degree, and 81% had some college or a college degree.

Down Syndrome (DS)—This group included 34 boys ranging in chronological age from 4.5 years to 15.9 years (M = 9.7 years), with a mean nonverbal cognitive age equivalent of 5.0 years on the *Leiter-R*. Parental report confirmed that the source of DS was Trisomy 21 for 32 of the boys and translocation for one boy. Eighty-eight percent of the participants were Caucasian, and 12% were African American. Nine percent of the mothers for this group of participants had a terminal education level of a high school degree, and 91% had some college or a college degree.

Typically developing (TD) boys—This group included 45 TD boys who were at similar nonverbal developmental ages as the boys with FXS and DS. The TD boys, ranging in age from 2.8 years to 7.8 years (M = 5.0 years), had a mean nonverbal cognitive age equivalent of 5.2 years on the *Leiter-R*. The TD boys were recruited from pediatricians' offices, childcare centers, and schools in North Carolina. Seventy-one percent of the boys were Caucasian, 18% were African American, and 11% reported another ethnicity. All but one of the mothers of the participants in this group had some college or a college degree.

Many of the participants in the present study were also participants in the Roberts and colleagues (2005) study of single-word phonological skills in these groups. Of the present

participants, 17 boys with FXS-O, 23 boys with FXS-ASD, 22 boys with DS, and 26 TD boys were participants in the previous study.

Assessment of Cognitive Skills

The Brief IQ composite from the *Leiter International Performance Scale - Revised* (*Leiter-R*) was administered as a measure of nonverbal cognition. This scale measures nonverbal cognitive abilities by assessing spatial reasoning, sequencing, and patterning skills. Item reliability and validity have been published for this standardized test (Roid & Miller, 1997). An age equivalent score was calculated for each participant.

Determination of Autism Status

The ADOS was administered to all participants with FXS to determine autism status at the first assessment of a larger longitudinal study. The ADOS is a standardized scale using observation of children's communicative and social behaviors to discriminate autism spectrum disorder from other developmental disorders and normal behavior. The ADOS yields categorical scores of "no autism," "spectrum", and "autism" based on three subscores for *Communication, Social Interaction,* and *Communication + Social Interaction*. To be included in the "no autism" group, a participant had to score less than 7 for Modules 1 and 3 or less than 8 for Module 2 and score below the spectrum cutoff on at least one of the three subscores. In order to be included in the "spectrum" group, a participant had to score in the range of 7 to 11 for Module 1, 8 to 11 for Module 2, or 7 to 9 for Module 3, and score at or above the spectrum cut-off score for each of the three subscores. In order to be included in the "autism" group, a participant had to score 12 and above for Modules 1 and 2 or 10 and above for Module 3 and score at or above the autism cut-off score for each of the three subscores. Examiners provided connected and behavioral cues or "presses" in semi-structured activities to allow the child opportunities to exhibit behaviors characteristic of autism. Trained examiners scored videotapes of ADOS interactions, and reliability computed on 16% of the boys was .93 on diagnosis (range .81 to 1.00). A total of 32 boys with FXS received an ADOS score of "no autism," and 31 received a score of "spectrum" or "autism." In our analyses, boys with FXS with scores of spectrum or autism were combined into a single group (FXS-ASD). The boys in the FXS-ASD group received a mean ADOS score of 10.8 (range 7 to 19), while the boys with FXS-O had a mean ADOS score of 4.7 (range 0 to 11). Because ASD is a static diagnosis that should not change over time, it was not necessary that the ADOS scored for diagnosis of ASD be administered at the same assessment at which the speech sample was collected. The speech sample was collected at the same assessment time point as the ADOS for 17 of the boys with FXS, approximately one year later for 1 of the boys, approximately 2 years later for 21 of the boys, and approximately 3 or more years later for 24 of the boys.

Phonological Assessment

Spontaneous speech samples were collected for all study participants using the ADOS. The ADOS is a semi-structured play assessment in which the evaluator elicits social and adaptive behaviors for the diagnosis of ASD. The approximately 45 minute assessment includes several developmentally appropriate conversational presses and toy-based interactions. One of the three modules of the ADOS was administered based on each child's developmental and language proficiency. Module 1 was administered to boys using single word utterances and simple phrases, Module 2 was administered to boys using connected speech ranging from three-word phrases to verbal fluency, and Module 3 was administered to the older and adolescent boys who were verbally fluent (Lord et al., 2001). For the speech samples elicited using Module 1 included free play, a pretend birthday party, a snack break, and interactions using bubbles and a balloon. Speech samples elicited using Module 2 or 3 included make-believe play with toys, joint interactive play, a book activity, pretend birthday party, snack, and bubbles and/or

balloon interactions. The ADOS was administered in its entirety to diagnose ASD, and the portion required to collect 100 first occurrence words for each participant was transcribed as the participant's connected speech sample. These connected speech samples yielded mean numbers of words per utterance of 3.5 for the FXS-O group, 3.4 for FXS-ASD, 2.7 for DS, and 4.3 for the TD group. All speech samples were audiotaped using a portable Digital Auditory Tape (DAT) TASCAM (DA-P1) recorder with a Shure WBH 53 headset microphone system and videotaped using a Sony (DCR-TVR27) Digital 8 Camcorder. Using guidelines for connected speech transcription outlined in the *Programs to Examine Phonetic and Phonologic Evaluation Records* (PEPPER) manual (Shriberg, 1986), the participants' speech samples were glossed by a trained speech-language pathologist (SLP) until a minimum of 100 intelligible first-occurrence words were obtained. Samples were glossed by utterance, with all unintelligible words marked using one 'x' per syllable. Partially intelligible utterances were transcribed. Speech produced while the participant was singing or book reading was not considered spontaneous speech and therefore was not glossed for transcription. Any speech sample that contained fewer than 100 intelligible first-occurrence words was dropped from the dataset. One sample from the FXS-O group, 5 from the FXS-ASD group, 6 from the DS group, and 0 from the TD group were dropped for this reason. In order to reduce any effects of inconsistent or poor sound recording quality, a second SLP verified the glosses for each speech sample via DAT recording, and disagreement on the glossing of any utterance was resolved by consensus. The gloss was then verified a second time by video and any previously unintelligible utterances that were intelligible with a visual context were added to the transcript. Gloss reliability was assessed using word-by-word comparison of two glosses of the same speech sample completed by two trained glossers. Gloss reliability was calculated for at least 10% of each diagnostic group and judged to be adequate at 81.6%.

After glossing was verified, the target transcription was written below the gloss of each utterance. A third SLP then listened to each speech sample and was permitted to replay any portion of the speech sample up to 3 times according to Shriberg and colleagues' procedures for phonetic transcription (Shriberg, Kwiatkowski, & Hoffman, 1984). This transcriber phonetically transcribed each utterance of the speech sample using narrow transcription guidelines, as described in Shriberg and Kent (2003), only marking speech productions that were different from the target transcription. Three word types included in the gloss transcriptions but excluded from the analyses of phonological accuracy were interjections (e.g., *ah, oh, yay, whoops*), symbolic noises that have conventional spellings (e.g., *achoo, choo-choo, boing, vroom*), and single words used as affirmative or negative responses (*yes, yeah, yep, no, nope, okay, uhhuh*).

Inter-observer agreement for broad transcription was computed on at least 10% of the speech samples from each of the four diagnostic groups (4 boys with FXS-O, 4 boys with FXS-ASD, 4 boys with DS, and 5 TD boys). Point-by-point comparison of broad transcription of connected phonological units was made such that each segment of an utterance had to be identical to count as inter-observer agreement. The average percentage agreement between two transcribers for broad transcription was 89.1% (range from 80.6% to 98.2%; 87.8 % for FXS-O, 87.8% for FXS-ASD, 86.1% for DS, and 93.5% for TD). For narrow transcription, the average percentage agreement between two transcribers was 87.2% (range from 76.2% to 98.0%; 86.4% for FXS-O, 86.3% for FXS-ASD, 82.6% for DS, and 92.4% for TD).

Phonological Assessment in Connected Speech

Phonological assessment of the connected speech samples included measures of consonant production accuracy, phonological process occurrence, and percentage of intelligible words. All of these measures were computed by Computerized Profiling (Long, Fey, & Channell, 2003).

Accuracy of consonant production—The accuracy of consonant production was measured by calculating the percentage of consonants correct (PCC; Shriberg & Kwiatkowski, 1982), which is the total number of correctly produced consonants divided by the total number of consonant targets. PCC has been found to be correlated with speech intelligibility in conversation and is a good index of speech disorder severity (Shriberg, Austin, Lewis, McSweeny, & Wilson, 1997).

Proportion of whole word proximity—The accuracy of whole word production in connected speech was measured using the calculation of Proportion of Whole Word Proximity (PWP; Ingram, 2002). PWP provides a comprehensive phonological analysis of an entire word by considering the accuracy of the production of all segments in a word, thereby taking length and complexity of the production into consideration. PWP can also be used as an indirect measure of speech intelligibility in connected speech (Bernthal & Bankson, 2004; Ingram, 2002). PWP is calculated by adding the number of all segments in a word and the number of correctly produced consonants in that word then dividing this number by the total number of segments plus the number of consonants in the target word. For example, the production of the target word *swim* (4 segments + 3 consonants = 7) as /sIm/ (3 segments + 2 correct consonants = 5) yields a PWP of 5/7 = .71.

Phonological process occurrence—Phonological processes, systematic sound changes that children adopt to simplify speech, can affect an entire class of sounds (e.g., fricatives), a particular sequence of sounds (e.g., st- blends), or the syllable structure of words (Bernthal & Bankson, 2004). We analyzed the presence of normally occurring processes for each group of participants. Each of these normally occurring processes fell into one of three categories: syllable structure, substitution, or assimilation processes (Grunwell, 1987). Because assimilation processes occurred rarely in boys with FXS in the present study and in the examination of phonological process occurrence in single words by Roberts and colleagues (2005), we limited our investigation to syllable structure and substitution processes in the current study.

For each process, the number of occurrences of each process was divided by the number of opportunities for that process, and this quotient yielded the percentage of process occurrence for each category. An overall percentage of process occurrence was computed for both of the phonological process categories by averaging the percentages of the individual phonological processes composing the two categories.

Percent Intelligible Words

To measure connected speech intelligibility, we used an objective measure of connected speech intelligibility, the percentage of intelligible words (PIW), which calculates the number of words understood by the listener divided by the total number of target words in the gloss transcript (Gordon-Brannan & Hodson, 2000; Shriberg, & Kwiatkowski, 1985). Because the ratio of monosyllabic words to polysyllabic words has been reported as approximately 3 to 1 in a typical speech sample (Shriberg & Kwiatkowski, 1980, 1983), we used this ratio to estimate the number of unintelligible words in a string of unintelligible symbols per the guidelines described in the PEPPER user's manual (Shriberg, 1986). According to this procedure, the first three unintelligible syllables are marked as monosyllabic words, and the next two syllables are marked as a two-syllable word. This convention is repeated to the end of the string of unintelligible syllables.

Analysis Strategy

Data analyses were completed to determine whether there were group differences on five outcomes: PCC, percent occurrence of syllable structure processes, percent occurrence of

substitution processes, PWP, and PIW. To test for between group differences on these measures of connected speech production, several analyses were completed. First, three phonological accuracy variables, PCC, PWP, and PIW were assessed in separate univariate general linear models (GLM). Second, a series of GLMs were run on the two phonological process variables: syllable structure processes and substitution processes. In all of the above models, the dependent variable was assessed as a function of diagnostic group (FXS-O, FXS-ASD, DS, and TD), nonverbal mental age (as measured by the *Leiter* Brief IQ) and their interaction. The purpose of including mental age is to control for the possibility of between group differences in cognitive development. Effect sizes for significant differences between groups were computed using the formula for Cohen *d*. Effect sizes were computed to illustrate the magnitude of group differences on each of the dependent variables, with a Cohen *d* of .2 designated as small, .5 as medium, and .8 as large (Cohen, 1988).

Results

Phonological Accuracy

Percent Consonants Correct—A significant main effect was found by diagnostic group, $F(3, 134) = 38.73$, $p < .0001$, for phonological accuracy as measured by PCC. The boys with DS were significantly different from all of the other groups, having a lower PCC (71.6%) than the boys with FXS-O (90.9%), the boys with FXS-ASD (88.2%), and the TD boys (89.7%). Post hoc analyses indicated that compared to the boys with DS, the effect sizes were large for the boys with FXS-O ($d = 2.22$), the boys with FXS-ASD ($d = 2.02$), and the TD boys ($d = 2.14$). The boys with FXS-O and FXS-ASD were not significantly different from each other ($d = .20$) or the TD boys ($d = .08$ and .12, respectively) in PCC.

Proportion of Whole Word Proximity—A significant main effect was found by diagnostic group, $F(3, 134) = 33.45$, $p < .0001$, for phonological accuracy as measured by PWP. The boys with FXS-O were not significantly different from the boys with FXS-ASD or the TD boys in PWP. The boys with DS were significantly different from the other groups, having a lower PWP (86.4) than the boys with FXS-O (95.5), the boys with FXS-ASD (94.1), and the TD boys (95.4). Post hoc analyses indicated that as compared to the boys with DS, the effect sizes were again large for the boys with FXS-O ($d = 2.04$), the boys with FXS-ASD ($d = 1.77$), and the TD boys ($d = 2.05$). The boys with FXS-O and FXS-ASD were not significantly different from each other ($d = .27$) or the TD boys ($d = .01$ and .27, respectively) for PWP.

Phonological Processes

The results of the multivariate model indicated that the effects for diagnostic group were significant, $F(3, 131) = 14.84$, $p < .001$, but that neither *Leiter-R* nor its interaction with group was significant. The following univariate models probe the group effects for syllable structure processes and substitution processes.

Syllable structure processes—GLM was used to determine if significant differences existed across diagnostic groups for the percentage of syllable structure processes. The GLM produced a significant main effect by diagnostic group, $F(3, 131) = 14.98$, $p < .0001$). The boys with FXS-O, the boys with FXS-ASD, and the TD boys did not differ in their percentage of occurrence of syllable structure processes (2.1%, 2.6%, and 1.5%, respectively). The boys with DS used syllable structure processes significantly more often (6.1%) than all other groups. Post hoc analysis indicated that as compared to the boys with DS, the effect sizes were large for the boys with FXS-O ($d = 1.22$), the boys with FXS-ASD ($d = 1.12$), and TD boys ($d = 1.49$). The boys with FXS-O and FXS-ASD were not significantly different from each other ($d = .10$) or the TD boys ($d = .27$ and .36, respectively) in syllable structure process occurrence.

Substitution processes—The GLM produced a significant main effect for diagnostic group ($F(3, 131) = 8.93$, $p < .0001$). The boys with FXS-O did not differ from the boys with FXS-ASD or the TD boys in the percentage of occurrence of substitution processes (4.9%, 8.3%, and 6.5%, respectively). In the boys with DS, substitution processes occurred significantly more often (13.2%) than in the other groups. Post hoc analysis indicated that compared to the boys with DS, the effect sizes were large for the boys with FXS-O ($d = 1.23$), the boys with FXS-ASD ($d = .76$), and TD boys ($d = .95$). The boys with FXS-O and FXS-ASD were not significantly different from each other ($d = .46$) or the TD boys ($d = .28$ and .19, respectively) in substitution process occurrence.

Individual phonological processes—The individual phonological processes that comprise the syllable structure and substitution processes were examined (see Table 3 for percentage occurrence of each process). The most commonly occurring syllable structure process in the boys with FXS-O was cluster reduction and the most commonly occurring substitution processes were fricative simplification and deaffrication. Similarly, in the boys with FXS-ASD, the most commonly occurring syllable structure process was cluster reduction and the most commonly occurring substitution process was cluster simplification, followed by liquid simplification, deaffrication, and fricative simplification. In the boys with DS, the most commonly occurring syllable structure process was cluster reduction, and the most commonly occurring substitution process was cluster simplification, followed liquid simplification, palatal fronting, fricative simplification, later stopping, and deaffrication. In the TD boys, cluster reduction was the most commonly occurring syllable structure process and the most commonly occurring substitution process was later stopping, followed by liquid simplification and cluster simplification. All other syllable structure and substitution processes occurred relatively infrequently among the four groups. None of the individual assimilation processes had an occurrence of more than 1% for any of the groups.

Intelligibility

Percent Intelligible Words—A significant main effect was found by diagnostic group, $F(3, 134) = 25.72$, $p < .0001$) for the measure of PIW. The PIW scores of the boys with FXS-O (82.0%), the boys with FXS-ASD (81.7%), and the boys with DS (81.2%) were significantly lower than those of the TD boys (95.9%). Post hoc analysis indicated that as compared to the TD boys, the effect sizes were large for the boys with FXS-O ($d = 1.61$), the boys with FXS-ASD ($d = 1.61$), and boys with DS ($d = 1.55$). The boys with FXS-O and FXS-ASD were not significantly different from each other ($d = .01$) or the boys with DS ($d = .06$ and .06, respectively).

Discussion

The current study provides new information about phonological accuracy and intelligibility in the connected speech of boys with FXS and DS. We found that the boys with FXS (both with and without ASD) did not differ on measures of phonological accuracy or phonological process occurrence but scored lower on a measure of speech intelligibility when compared to the TD boys. Both groups of boys with FXS scored higher on phonological accuracy, scored lower on phonological process occurrence, and were not significantly different from the boys with DS in intelligibility. The boys with DS scored lower on all measures of phonological accuracy and speech intelligibility and higher in phonological process occurrence than the TD boys, indicating that they exhibit greater delays in phonological development relative to the boys with FXS and younger TD boys after controlling for nonverbal mental age.

The connected speech patterns of boys with FXS were similar to those of younger TD boys at a similar nonverbal mental age. The boys with FXS exhibited similar mastery of syllable and

word shapes as measured by Proportion of Whole Proximity scores, which were almost identical to those of the TD boys, and both groups of boys with FXS used the same phonological processes as the TD boys with similar percentages of occurrence. Though the average Percent Consonants Correct, Proportion of Whole Word Proximity, and phonological process occurrence scores were similar for the boys with FXS and the TD boys, it is important to point out that there was great individual variability among the participants with FXS. For example, one 13-year-old boy with FXS-O had a Percent Consonants Correct score of 74, a Proportion of Whole-Word Proximity score of 88, and a percent occurrence of substitution processes nine, whereas another 13-year-old boy with FXS-O had Percent Consonants Correct and Proportion of Whole Word Proximity scores of 100 and a zero percent occurrence of substitution processes.

Our finding that the boys with FXS exhibited phonological accuracy in connected speech similar to that of younger TD boys is consistent with previous studies documenting the phonological accuracy in FXS at the word level. Three previous studies found that boys with FXS (ages 3 to 34 years) exhibited common sound substitutions, omissions, and distortions developmentally appropriate for younger TD children (Hanson et al. 1986; Madison et al., 1986; Prouty et al., 1988). The present results are also in agreement with a study by Paul, Cohen, and colleagues (1984), in which three boys with FXS (ages 10 to 13) were observed to use common phonological processes such as liquid simplification and final consonant deletion, two of the most commonly occurring processes in the boys with FXS in the present study. The present data regarding phonological accuracy in connected speech are also consistent with those of single-word speech samples obtained by Roberts and colleagues (2005), many of which are from the same participants as those in the present study. In connected speech, like single words, the boys with FXS exhibited a delay in phoneme acquisition relative to same-age TD peers, with a Percent Consonants Correct score similar to that of the younger TD boys. We also found that the boys with FXS had a similar Proportion of Whole-Word Proximity in connected speech as in single words. Finally, the boys with FXS used many of the same phonological processes in connected speech that Roberts and colleagues (2005) found to be productive at the single word level, such as final consonant deletion, cluster reduction, palatal fronting, later stopping, liquid simplification, and cluster simplification as well as infrequently used processes, such as final consonant deletion, early stopping, and voicing/devoicing errors.

One difference between the present results and some previous reports is in the finding of speech intelligibility in FXS (Madison et al., 1986; Paul, Cohen, et al., 1984; Spinelli et al., 1995). Madison and colleagues (1986) and Paul, Cohen, and colleagues (1984) reported that despite the occurrence of phonological errors and phonological processes, boys with FXS were intelligible at the single word level, but the boys with FXS in the present study were found to be less intelligible in connected speech than the TD boys. Our findings regarding reduced intelligibility in FXS relative to the TD boys is consistent with another study, in which intelligibility is reported to be reduced in this population. Spinelli and colleagues (1995) studied eight males with FXS (ages 6 to 26 years) and found that listeners had difficulty understanding all eight males as utterance length increased.

The finding of decreased intelligibility in FXS relative to TD peers despite similar scores on all measures of phonological accuracy suggests that the reduced speech intelligibility in children with FXS may not be due to differences in phonological accuracy but perhaps due to other connected factors such as prosody (e.g., rate, intonation) and fluency. These are two aspects of speech production in which boys with FXS are reported to differ from TD peers (Borghgraef, Fryns, Dielkens, Pyck, & Van den Bergh, 1987; Hanson et al., 1986; Palmer et al., 1988; Reiss & Freund, 1992; Spinelli et al., 1995), but whether boys with FXS differ in prosody from TD peers requires further study.

The finding that the boys with FXS-ASD and the boys with FXS-O did not differ in phonological accuracy is supported by a previous study of boys with co-morbid FXS and ASD (Roberts et al., 2007). Roberts and colleagues (2005) compared 49 boys with FXS-ASD and 33 boys with FXS-O and found that the groups did not differ in phonological accuracy at the single word level when at similar nonverbal cognitive levels. In other studies, phonological accuracy was not found to be delayed in individualswith autism beyond mental age expectations (Bartolucci & Pierce, 1977; Kjelgaard & Tager-Flusberg, 2001; Rice et al., 2005; Shriberg et al., 2001). For example, Kjelgaard & Tager-Flusberg studied 89 children with autism (ages 4 to 14 years) and found that expressive phonology at the one word level was in the normal range for chronological age (Kjelgaard & Tager-Flusberg, 2001). However, our findings differed with another study, which found that children with autism exhibited more motor speech and speech production difficulty than TD children (Adams, 1998). Another finding of the present study was that boys with FXS-ASD had similar phonological accuracy when compared to younger TD boys. This finding is supported by a previous study by Shriberg and colleagues, in which they found that 30 males with autism (ages 10 to 50 years) had more residual articulation errors than their TD same-age peers, suggesting that their speech production was similar to younger TD peers (Shriberg, 2001).

The connected speech measures of boys with DS revealed delays compared to the younger TD boys with a similar nonverbal mental age on all measures. The boys with DS had a lower Percent Consonants Correct score (86) than the younger TD boys (95), and had a lower average Proportion of Whole Word Proximity score of 86 as compared to that of the TD boys (95). The boys with DS used many of the same phonological processes as the TD boys, such as later stopping, liquid simplification, and cluster simplification, yet these processes occurred more often for the boys with DS than the TD boys. Thus, the boys with DS made more errors in consonant production and were more likely to change syllable shapes by omitting segments or syllables in a word than the TD boys. This change in word shapes (i.e., reduction of clusters, omission of phonemes, omission of syllables) that results in the occurrence of syllable structure processes can have a significant impact on intelligibility (Hodson & Paden, 1991). There also was great individual variability among the participants with DS in all of these measures. For example, one 6-year-old boy with DS had a Percent Consonants Correct score of 56 and a Proportion of Whole Word Proximity score of 76; he also used syllable structure processes in 13% of opportunities. In comparison, another 6-year-old boy with DS had a Percent Consonants Correct score of 89 and a Proportion of Whole Word Proximity score of 95; he also used syllable structure processes in only 1% of opportunities.

The current findings regarding phonological accuracy in connected speech in DS are consistent with those at the single-word level as reported by Roberts and colleagues (2005). In single words as in connected speech, the boys with DS exhibited a delay in phonemic acquisition (as measured by Percent Consonants Correct and Proportion of Whole Word Proximity) relative to same-age TD peers. Phonological process occurrence was similar in single words and connected speech as well. Except for early stopping, all of the phonological processes Roberts and colleagues found to be productive at the single word level occurred often in connected speech, including final consonant deletion, cluster reduction, palatal fronting, later stopping, liquid simplification, and cluster simplification.

The current findings that boys with DS are delayed in their phonological development beyond mental age expectations when compared to their TD peers is supported by previous studies in which children with DS exhibited later phoneme emergence and suppressed common developmental phonological processes at a slower rate when compared to TD children but were considered delayed, not different, in their phonological development from TD peers (Bleile & Schwarz, 1984; Kumin et al., 1994; Smith & Stoel-Gammon, 1983; Stoel-Gammon, 1980). Smith and Stoel-Gammon (1983) found that compared to four TD children (ages 1 to 3 years),

five children with DS (ages 3 to 6) exhibited delayed phonemic acquisition and the occurrence of similar phonological processes in single words relative to the younger TD children.

In the present study, the boys with FXS and DS scored differently on all speech measures except that of intelligibility. The boys with FXS scored higher on measures of phonological accuracy, Percent Consonants Correct, and Proportion of Whole Word Proximity and had a lower occurrence of syllable structure and substitution processes than the boys with DS, but they did not score differently on Percent Intelligible Words. The same phonological processes occurred in the boys with DS and the boys with FXS but occurred with more frequency in the boys with DS. It is possible that the cause of reduced intelligibility in FXS may be related to factors other than phonological accuracy or process usage, which may help explain why their intelligibility scores did not differ from the boys with DS despite scoring higher on all measures of phonological accuracy. Some possible causes for this intelligibility deficit in FXS may be related to suprasegmental characteristics or fluency deficits exhibited by these boys. Rapid and fluctuating rate, intonation differences, stuttering, or cluttering which have been described as occurring in some males with FXS (Borghgraef, Fryns, Dielkens, Pyck, & Van den Bergh, 1987; Brun-Gasca and Antigas-Dallares, 2001; Hanson, Jackson, & Hagerman, 1986; Reis & Freund, 1992) may possibly contribute to intelligibility in connected speech without affecting phonological production. Other explanations may be related to acoustic characteristics of the connected speech signal produced by these children that are too subtle to be measured subjectively through phonetic transcription such as atypical pauses or stress or rate fluctuations.

The present study has several limitations that should be considered. First, percentage of intelligibility is relatively high for all four groups of participants (greater than 80%). This may be due in part to our method, in which each highly trained glosser or transcriber was allowed to listen to an utterance up to three times, and audio glosses were verified using video. Our familiarity with the play materials used and the ability to use contextual clues from the video to gloss single words, rather than utterances, also may have yielded a higher intelligibility score than a naïve listener would have given the speech sample as a whole. Given the varying amount of unintelligible utterances in the connected speech samples, it is also possible that applying a convention to “estimate” the Percent Intelligible Words inflated the outcome in the boys with FXS and DS, who had the most unintelligible utterances. This is a limitation of assessing connected speech in children who are not completely intelligible, but as the same procedure was used to calculate intelligibility for all participants, differences between groups should be preserved. A second limitation of the current study is that the standard deviations for the occurrence of phonological processes were quite large, indicating wide variability in the percentage of occurrence within each group. This may be due in part to the variable nature of spontaneous speech samples, another limitation of assessing connected speech, as children using less complex syllable shapes and fewer types of words may have fewer opportunities to use many individual phonological processes than children using more complex syllable shapes. A third limitation of the study, similar to that of the single-word study conducted by Roberts et al. (2005), only phonemic consonants and known target words were studied. We were therefore unable to describe the types of errors made during unintelligible speech in which target phonemes were unknown, and we did not study vowel accuracy or suprasegmental and nonsegmental aspects of speech, such as speaking rate, pauses, or fluency, and how they might impact speech production errors. A fourth limitation of the study is that comparisons of boys with FXS and DS were not sufficient for determining syndrome-specific speech characteristics; future studies should compare these children with other syndrome groups on these speech measures. A fifth and final study limitation is that our analysis represented the phonological accuracy and intelligibility characteristics of our participants at only one time point; studying the developmental trajectories of our participants may provide more information about the differences in the rate and patterns of speech development in these populations.

These limitations have implications for future research directions in connected speech phonological accuracy for these populations. First, future research should address the concurrent suprasegmental features of connected speech, such as prosody, rate, pauses, and fluency, and their possible effects on phonological accuracy and speech intelligibility. Second, future studies should address phonological development across several time points, revealing the patterns of development and developmental trajectories of each of our participant groups. Finally, our findings do not suggest why children with FXS are less intelligible than their TD counterparts despite almost identical phonological outcomes. Future research should focus on identifying possible causal factors that may contribute to limited intelligibility, such as suprasegmental characteristics, fluency, and more objective acoustic analyses of speech production.

The results of the present study have important clinical implications regarding assessment and intervention in the phonological development of boys with FXS and boys with DS. First, because there are considerable differences among individuals and both populations have speech delays relative to chronological age, a comprehensive speech assessment focusing on the child's phonemic inventory, word shapes, phonological process occurrence, and intelligibility in connected speech should be completed. Because both groups exhibited similar phonological accuracy in words as compared to connected speech, a single word articulation test may be an efficient and effective method of assessing phonological skills in boys with FXS and DS. In boys with FXS however, a comprehensive evaluation should also include a measure of connected speech intelligibility, since intelligibility in this group is reduced compared to TD peers despite age-appropriate phonological accuracy scores. Because phonological accuracy cannot explain the reduced intelligibility in FXS, it is important to assess other factors that could affect speech intelligibility, such as oral motor speech skills, rate, fluency, and prosody in this group. Similar to assessment, intervention approaches may differ for children with FXS and DS since their speech production accuracy and phonological process occurrence differs. Because the phonological errors and phonological process usage displayed by the boys with FXS are similar to those displayed by younger TD children, intervention approaches that have proven successful in improving speech production errors in children with IQs in the normal range may be utilized. The boys with FXS showed delayed phonemic acquisition but adequate retention of word shapes as compared to the TD boys, therefore therapy should focus on correcting individual phoneme articulation and suppressing substitution processes as well as considering other speech aspects, such as prosody, that may be limiting their intelligibility in connected speech. Intervention in boys with DS should focus on improving not only intelligibility, which was also significantly lower than that of the TD boys, but also increasing phoneme acquisition, suppressing commonly used phonological processes, and retaining word shapes. The cycles approach may be particularly effective in suppressing syllable structure and substitution processes in both populations (Hodson, 2006b; Hodson & Paden,1991). The complexity approach (Gierut, 2001, 2005), which is highly structured and targets more complex sounds than easier sounds, may also be useful in increasing the phonetic repertoire of individuals with FXS and DS. The boys with DS, in particular, may benefit from therapy tasks that address "syllableness," in which the retention of target word shapes is addressed by marking often-deleted syllables and segments in blends and words (Hodson & Paden, 1991).

Acknowledgements

Research was supported by the National Institute of Child Health and Human Development (1 R01 HD38819, 1 R01 HD044935, and 1 R03 HD40640) and the March of Dimes. We would like to thank the boys who participated in our study as well as their families. We would also like to acknowledge the assistance of the data collectors on the Carolina Communication Project, Ms. Anne Edwards, Ms. Lauren Moskowitz, Ms, Ms. Cheryl Malkin, Ms. Joy Scott, and Dr. Beth Hennon. We thank Ms. Jan Misenheimer and Lauren Nelson for data analysis.

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Table 1
Chronological and Developmental Age Levels by Group

Age	FXS-O (N=32)	FXS-A (N=31)	DS (N=34)	TD (N=45)
Chronological Age (in years)				
M	10.9	10.1	9.7	5.0
SD	2.6	3.1	2.9	1.1
Range	3.2 - 14.5	5.0 - 15.4	4.5 - 16.0	2.8 - 7.8
Leiter-R Developmental Age (in years)				
M	5.3	5.1	5.0	5.2
SD	0.8	0.8	1.0	1.3
Range	2.2 - 6.6	2.4 - 6.6	3.1 - 8.2	3.2 - 10.3

Table 2
Means (standard deviations) of Summary Variables by Group

	FXS - O (N = 32)	FXS - ASD (N = 31)	DS (N = 34)	TD (N = 45)
	Mean (SD)	Mean (SD)	Mean (SD)	Mean (SD)
Phonological Accuracy				
% Consonants Correct	90.9 (3.01)	88.2 (6.03)	71.6 (9.57)	89.7 (8.64)
Prop. Whole Word Proximity	95.5 (2.94)	94.1 (3.18)	86.4 (5.24)	95.4 (3.78)
Intelligibility				
% Intelligible Words	82.0 (10.71)	81.7 (11.30)	81.2 (11.73)	95.9 (5.15)
Phonological Processes				
% Syllable Structure	2.1 (1.73)	2.6 (2.13)	6.1 (4.43)	1.5 (2.06)
% Substitution	4.9 (4.04)	8.3 (6.12)	13.2 (7.42)	6.5 (6.82)
% Assimilation	0.3 (0.51)	0.3 (0.71)	1.1 (1.73)	0.1 (0.55)

Table 3

Means (standard deviations) of Individual Phonological Process Usage by Group

	FXS - O (N = 32)	FXS - ASD (N = 31)	DS (N = 34)	TD (N = 45)
Phonological Process	**Mean (SD)**	**Mean (SD)**	**Mean (SD)**	**Mean (SD)**
Syllable Structure				
% Final Consonant Deletion	2.0 (2.19)	3.5 (3.96)	8.2 (11.03)	0.8 (1.28)
% Cluster Reduction	8.2 (6.99)	9.1 (7.56)	20.8 (13.09)	5.8 (8.54)
Substitution				
% Palatal Fronting	6.6 (7.91)	8.1 (13.14)	19.9 (25.66)	6.1 (12.03)
% Later Stopping	3.9 (5.96)	5.8 (7.50)	15.7 (15.34)	15.8 (18.33)
% Liquid Simplification	6.4 (12.66)	17.0 (23.52)	20.8 (20.08)	13.9 (23.38)
% Cluster Simplification	8.2 (14.99)	20.6 (25.79)	23.4 (25.53)	9.8 (18.51)
% Fricative Simplification	10.2 (13.92)	10.6 (18.59)	16.5 (22.54)	2.4 (4.36)
% Deaffrication	9.9 (13.62)	11.7 (16.91)	15.7 (25.15)	2.6 (7.97)
Assimilation				
% Reduplication	0 (0.00)	0 (0.00)	0.2 (0.61)	0.1 (0.53)
% Velar Assimilation	0.5 (1.02)	0.1 (0.36)	0.9 (1.99)	0.1 (0.41)
% Nasal Assimilation	0.1 (0.25)	0.5 (1.39)	1.2 (2.42)	0.2 (0.93)

Table 4

Adjusted Means (adjusted for Leiter Developmental Age) and Analysis Results Comparing the Groups on the Phonological Measure Summary Variables

	Group F	FXS-O M (SE) N=32	FXS-ASD M (SE) N=31	DS M (SE) N=34	TD M (SE) N=45
Percent Consonants Correct	Overall test F(3,134) 38.73	89.8[a] (1.57)	88.2[a] (1.53)	71.6[b] (1.46)	89.2[a] (1.25)
Proportion of Whole Word Proximity	F(3, 134) 33.45	95.0[a] (.80)	93.9[a] (.78)	86.5[b] (.75)	95.0[a] (.63)
Percent Intelligible Words	F(1, 134) 25.72	80.8[a] (1.80)	80.8[a] (1.79)	81.4[a] (1.71)	95.9[b] (1.46)
Syllable Structure Processes	F(3,131) 14.98	2.6[a] (.60)	2.9[a] (.62)	6.3[b] (.56)	1.8[a] (.48)
Substitution Processes	F(3,131) 8.93	4.9[a] (1.25)	7.9[a] (1.29)	12.8[b] (1.27)	6.8[a] (1.0)

Note. Letter superscript identifies statistically significant differences (if groups have the same letters, then differences were not significant)

***All $p < .001$