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Longitudinal Profiles of Expressive Vocabulary, Syntax, and Pragmatic Language in Boys with Fragile X Syndrome or Down Syndrome

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Abstract

Background—Fragile X syndrome (FXS) and Down syndrome (DS) are the two leading genetic causes of intellectual disability, and FXS is the most common known genetic condition associated with autism. Both FXS and DS are associated with significant language impairment, but little is known about expressive language across domains over time or the role of autism in language development in FXS.

Aims—To compare three domains of language production (vocabulary, syntax, and pragmatics) over time within and across groups of boys with FXS with and without autism spectrum disorder (FXS-ASD, FXS-O), boys with DS, and typically developing (TD) boys.

Methods & Procedures—Twenty-nine boys with FXS-O, 40 boys with FXS-ASD, 34 boys with DS, and 48 younger TD boys of similar nonverbal mental age living in the United States participated. The Antonyms, Syntax Construction, and Pragmatic Judgment subtests of the Comprehensive Assessment of Spoken Language were administered annually over three years.

Outcomes & Results—TD boys scored higher than all other groups on all three subtests, boys with FXS-O and FXS-ASD scored higher than boys with DS in Syntax Construction, and boys with FXS-O scored higher than boys with FXS-ASD in Pragmatic Judgment. Within-group patterns varied between groups. Overall the TD group showed significantly more change over time than all other groups.

Declaration of Interest
The authors report no conflicts of interest.

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Conclusions & Implications—Findings suggest that expressive language skills and growth across various domains are more impaired in boys with FXS and DS than would be expected based on nonverbal mental age, that for boys with DS syntax is more impaired than would be expected based on intellectual disability, and that autism status affects pragmatic language in boys with FXS. Findings suggest that language production across domains should be addressed during assessment and intervention for boys with FXS and boys with DS, with differing group profiles also suggesting potentially different areas of focus.

Introduction

Fragile X syndrome (FXS) is the leading known inherited cause of intellectual disability (ID; Dykens et al., 2000, Hagerman & Hagerman, 2002), and is associated with a complex phenotype including moderate or severe ID (Abbeduto & Chapman, 2005, Bennetto & Pennington, 2002), social anxiety (Bregman et al., 1988, Cordeiro et al., 2011, Hagerman, 2002), attentional deficits (Hooper et al., 2000, Wilding et al., 2002), and impaired language (Finestack et al., 2009, Abbeduto et al., 2007, Roberts et al., 2008). FXS is also the most common known genetic condition associated with autism (Hagerman, 2002), with about 20–50% of males meeting full criteria for autism and up to three-quarters meeting criteria for an autism spectrum disorder (ASD) on gold standard measures (Clifford et al., 2007, Hall et al., 2008, Kaufmann et al., 2004, Philofsky et al., 2004, Rogers et al., 2001). Because they possess an unaffected X chromosome, females with FXS are typically less severely affected than males (Hagerman, 2002, Loesch et al., 2002, Reiss & Dant, 2003).

Down syndrome (DS) is the most common known genetic cause of ID (CDC, 2006) and unlike FXS is not inherited (except in rare cases of translocations). An extra copy of chromosome 21 causes the vast majority of cases (Trisomy 21); as noted, translocation and mosaicism are less frequent causes. Moderate ID is present in most individuals with DS, although ability level varies from severe ID to average intelligence (Pueschel, 1995, Roizen, 2007). Impairment in verbal short-term memory is common (Jarrold & Baddeley, 2001, Laws, 2002), whereas visuo-spatial processing and perception (Fidler et al., 2006, Jarrold et al., 1999) along with social skills (Dykens et al., 2006, Fidler et al., 2006) are generally considered to be relative strengths.

Both FXS and DS are associated with significant language delay, with strong evidence that language in both populations is disproportionately impaired over general cognitive abilities and language production is more impaired than comprehension (Abbeduto et al., 2007, Roberts et al., 2008, Finestack et al., 2009, Roberts et al., 2001, Martin et al., 2009). In the case of FXS, there is some evidence that individuals with co-morbid autism tend to perform more poorly than do those with only FXS on global language measures (Bailey et al., 2001, Philofsky et al., 2004, Rogers et al., 2001). Language impairment, and pragmatic language difficulty in particular, is a core characteristic of autism (Landa, 2000, Tager-Flusberg et al., 2005). The overlap of FXS and autism thus raises questions about whether autism in the context of FXS may influence language development in ways that are different than in FXS only. In this longitudinal study, we examined language production across domains and over time in boys with FXS with and without ASD, boys with DS, and typically developing (TD) boys.

Language Production in FXS

Several studies have examined the structural language (vocabulary and syntax) skills of males with FXS, and findings are somewhat mixed. Expressive vocabulary in males with FXS was comparable to nonverbal mental age (MA) in one investigation (Roberts et al., 2007c) and delayed relative to MA according to two others (Roberts et al., 2007a, Sudhalter et al., 1991), perhaps due to the assessment method. Specifically, boys with FXS without

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ASD produced fewer different words in conversation than TD MA-matches in one study (Roberts et al., 2007c) but another study using the Expressive Vocabulary Test found no differences between boys with FXS only and TD controls (Roberts et al., 2007c). However, Roberts, Price, and colleagues did find that boys with both FXS and ASD differed from TD boys, suggesting that ASD in FXS may negatively impact vocabulary skills. Another recent study found that males with only FXS did not differ from a small sample of males with comorbid FXS and autism in lexical diversity measured from a spontaneous language sample (Kover & Abbeduto, 2010).

Recent studies also suggest that males with FXS have delayed expressive syntax relative to MA (Estigarribia et al., 2010, Price et al., 2008, Roberts et al., 2007a), although some older studies report contrary results (Ferrier et al., 1991, Scarborough et al., 1991). For example, Estigarribia and colleagues (2010) recently demonstrated that boys with FXS with and without ASD produced fewer noun- and verb-related grammatical morphemes in conversation than TD boys, after controlling for nonverbal MA, maternal education, and articulatory skill. Syntactic complexity of males with FXS did not differ by autism status in two other recent studies using natural language samples (Kover & Abbeduto, 2010, Price et al., 2008).

There is general consensus that pragmatic language is impaired in males with FXS beyond cognitive level. During conversation, males with FXS tend to produce more noncontingent (off-topic or tangential) language (Wolf-Schein et al., 1987, Sudhalter & Belser, 2001) and verbal perseveration (Roberts et al., 2007b, Wolf-Schein et al., 1987, Sudhalter et al., 1990) than either boys with DS or TD boys of similar nonverbal MA. In narration, boys with FXS include fewer story actions than do TD boys of similar nonverbal MA (Estigarribia et al., 2011). Moreover, boys with both FXS and ASD are more noncontingent during conversation than boys with FXS only (Roberts et al., 2007b), and ASD in boys with FXS may also negatively affect narrative skills (Estigarribia et al., 2011).

Language Production in DS

Compared with FXS, more research has been conducted on language in individuals with DS. On standardized assessments of expressive vocabulary, children with DS scored lower than MA-matched TD children in several studies (Caselli et al., 2008, Hick et al., 2005, Roberts et al., 2007c), and children and adolescents with DS also produced fewer total and fewer different words in conversation and narration in one study (Chapman et al., 1998). Considerable evidence points to syntax delays in individuals with DS that are beyond expectations for cognitive level, using either standardized tests or measures derived from conversational and narrative samples (e.g., Eadie et al., 2002, Chapman et al., 1998, Price et al., 2008), although MLU and syntactic complexity continue to grow into late adolescence and young adulthood in DS (Chapman et al., 2002, Thordadottir et al., 2002). Expressive syntax was also found to be more impaired than expressive vocabulary in one study (Laws & Bishop, 2003), further evidencing the particular challenge that syntax poses for individuals with DS.

Pragmatic language findings have been more mixed than those for structural language. Strengths of the DS pragmatic profile compared with TD children matched on developmental or language age include the use of a variety of communicative functions with the exception of requests (Beeghly et al., 1990, Coggins et al., 1983), the ability to maintain a topic of conversation (Beeghly et al., 1990, Tannock, 1988), and picture-supported storytelling (Boudreau & Chapman, 2000, Miles & Chapman, 2002). Difficulties compared with MAmatched TD children may involve topic initiation and elaboration (Tannock, 1988, Roberts et al., 2007b), providing clear messages (Abbeduto et al., 2006), and requesting clarification of unclear messages (Abbeduto et al., 2008).

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Summary and Current Study

There is growing evidence that language production in boys with FXS across the domains of vocabulary, syntax, and pragmatics is more impaired than would be expected based on nonverbal MA alone. Evidence to date also suggests that language impairments are more severe among individuals with both FXS and autism, yet little is known about specific language domains impacted and whether they are differentially affected by autism status, nor is it clear how autism might affect the course of language development in FXS. The evidence for vocabulary and syntax deficits beyond general cognitive level is considerable for individuals with DS, although pragmatic language findings have been mixed. The relatively small number of studies that have compared young individuals with FXS and DS found that those with FXS produce more complex syntax (Finestack & Abbeduto, 2010, Price et al., 2008) but also show more pragmatic language difficulties (Roberts et al., 2007b, Wolf-Schein et al., 1987, Sudhalter & Belser, 2001). Studies comparing language production across domains over time and within groups are lacking, especially for boys with FXS.

In the present investigation, we examined expressive vocabulary, syntax, and pragmatic language longitudinally in boys with FXS and boys with DS to determine how the two leading genetic causes of ID affect the course of language development across different language domains, in comparison to each other and younger TD boys of similar nonverbal MA. Boys with FXS both with and without ASD were included to increase understanding of the effects of autism status on language in boys with FXS. In addition to determining whether the language profile of boys with FXS differs by autism status or from that of boys with DS and TD boys, within-group comparisons in particular may have important clinical implications for identifying strengths and weaknesses. This study addressed the following aims: 1) to document similarities and differences between groups across the domains of vocabulary, syntax, and pragmatics, 2) to chart the developmental trajectories in these domains within and across groups, and 3) to examine the influence of autism on the language abilities of boys with FXS.

Methods

Participants

As part of a large-scale longitudinal study (Roberts et al., 2007c), boys with FXS and DS were recruited from schools, physicians' offices, genetic clinics, and developmental clinics in the Eastern United States, as well as through the Research Participant Registry Core of the Carolina Institute for Developmental Disabilities at the University of North Carolina at Chapel Hill (UNC). TD boys were recruited from childcare centers, physicians' offices, and schools in North Carolina. Research procedures were approved annually by the institutional review board at UNC.

As previously mentioned, because females with FXS tend to have less severe impairments than males (Hagerman, 2002, Loesch et al., 2002), participants included only boys. Upon enrollment, parents reported that all boys were producing 40 or more words and combining two or more words. English was the primary language spoken in the home for all children. At Time 1, groups showed similar distributions of non-verbal developmental ages on the Leiter International Performance Scale-Revied (Leiter-R; Roid and Miller 1997), as described below. All boys with FXS had a diagnosis of full mutation FXS confirmed by DNA analyses. Boys were excluded for having an average hearing threshold greater than 30 dB HL in the better ear, determined from a hearing screening across 500; 1,000; 2,000; and 4,000 Hz with a Grason Stadler GSI 16 or 17, or MAICO MA 40 audiometer. Also excluded were boys with DS and TD boys who showed signs of autism at screening, and those who

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subsequently scored in the "autism" or "autism spectrum" range on the Autism Diagnostic Observation Schedule (ADOS; Lord et al., 2001), described below. Data were collected in up to three waves for all groups. The mean number of visits for children per group was 2.52 (FXS-O), 2.36 (FXS-ASD), 2.06 (DS), and 2.18 (TD). See Table 1 for background characteristics of all participant groups.

Assessments

Testing occurred in a quiet area of a home, school, or the Frank Porter Graham Child Development Institute, lasting about 6 hours for the full assessment and including several breaks. All assessments were audio-recorded with a Digital Auditory Tape TASCAM (DA-P1) and video-recorded with a Sony Digital8 video camera (Model DCR-TVR27).

Language Production—The participants' expressive vocabulary, syntax, and pragmatic language skills were assessed with the Comprehensive Assessment of Spoken Language (CASL; Carrow-Woolfolk, 1999). The CASL is a standardized assessment designed for individuals ages 3 to 21, and measures language skills in the domains of lexical/semantic, syntactic, pragmatic, and suprasyntactic (e.g., non-literal language) knowledge. The Antonyms subtest measures lexical skills. For this test, the examiner says a stimulus word and the participant must respond with a single word that means the opposite of the test word. Syntactic knowledge was assessed with the Syntax Construction subtest, for which the examiner reads the test item while the participant looks at a related picture. The participant must respond with a grammatically and semantically appropriate word, phrase, or sentence. The Pragmatic Judgment subtest assesses knowledge of appropriate language for diverse social situations. The examiner reads a script together with a vignette that represents some aspect of everyday life requiring a response from the child. The Antonyms, Syntax Construction, and Pragmatic Judgment subtests are the "core tests" (i.e., deemed by the test developers to be the most representative of lexical/semantic, syntactic, and pragmatic ability, respectively) of expressive language for younger children, and so were the most appropriate given the range of mental ages of study participants. As reported in the examiner manual, the CASL was standardized on a national sample of 1,700 individuals and correlates strongly (.80) with the Oral and Written Language Scales (OWLS; Carrow-Woolfolk, 1995), another measure of language across categories. A lesser but significant correlation (.596) was also found between the CASL and the Test of Language Development—Primary, Third Edition (TOLD--P:3; Newcomer & Hammill, 1997) in a sample of school-age children with specific language impairment (Hoffman et al., 2011). For the current study, age equivalent scores for each subtest were computed according to published norms except when a child received a raw score of 0, which was treated as missing in the analysis.

Nonverbal mental age—Four subtests of the Brief IQ composite of the Leiter-R (Leiter-R; Roid & Miller, 1997) were administered to measure nonverbal cognition: Figure Ground (identifying figures or designs within a complex stimulus), Form Completion (recognizing an object from its fragmented parts), Sequential Order (selection of the next item in a logical sequence of items), and Repeated Patterns (supplying the missing portion of a repeated pattern). The Leiter-R was standardized on 1,719 typical individuals aged 2–20 years. The Brief IQ composite has a test-retest coefficient of .96, its four subtests have alpha reliability coefficients from .75 to .88, and it correlates strongly with other commonly used IQ tests. Age equivalent scores were computed for all children in the present study according to published norms.

Autism in FXS—Boys with FXS were separated into two groups by autism status: (1) FXS with ASD (FXS-ASD), and (2) FXS without ASD, or "FXS-Only" (FXS-O). Autism status was determined with the Autism Diagnostic Observation Schedule (ADOS; Lord et al.,

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2001), an examiner-child interaction of developmentally appropriate structured and semi-structured activities which provides a child with opportunities to exhibit diagnostic signs of autism. The ADOS classifies individuals as having “autism,” “spectrum,” and “no autism.” Trained examiners scored videotaped ADOS administrations, and autism classifications were based on the revised ADOS algorithms (Gotham et al., 2008, Gotham et al., 2007). Mean interrater reliability calculated on a subset (18%) of the sample was 86% for individual items, 86% for diagnostic algorithm items, and 90% for diagnosis. Thirty-one boys with FXS were classified as “autism,” 9 as “spectrum,” and 29 as “no autism.” The FXS-ASD group consisted of the 40 boys identified as either “autism” or “spectrum.”

Data Analysis Strategy

Between- and within-group (FXS-O, FXS-ASD, DS, and TD) differences were examined on each of the CASL subtests (Antonyms, Syntax Construction, and Pragmatic Judgment), as were between and within-group differences in change over time, using a hierarchical linear model (HLM) with subtest nested within chronological age (as the marker of change over time) which was nested within child. The interaction of diagnosis by time was included for differential development between groups. Random intercepts were included in the model, but all other effects were fixed. This enabled the testing of between subscale differences as well as interactions of subscale with diagnosis and time and the three-way interaction of subscale, time, and diagnosis. Nonverbal MA (Leiter-R) and maternal education were included in the model to adjust the estimated means. Prior to analysis, MA and chronological age were grand-mean centered to reduce collinearity and so that main effects would be estimated at the mean of both of these variables. This did mean that the data were centered near the upper end of the TD group. Group differences were not evaluated outside the range of the data. A planned follow-up HLM was conducted that compared groups on the Pragmatic Judgment subtest controlling for differences in scores on the Antonyms and Syntax Construction subtests in addition to nonverbal MA and maternal education. Controlling for structural language abilities is a more recent approach in group comparison studies of pragmatic language (Tager-Flusberg, 2004). Finally, recent research advances have made available a 10-point calibrated autism severity metric based on the revised ADOS algorithms as a method of quantifying ASD severity independent of age and verbal IQ (Gotham et al., 2009). Models were rerun on the FXS sample as a whole (combining FXS-O and FXS-ASD) with diagnosis replaced by autism severity.

Results

Unadjusted means and standard deviations for the three CASL subtests at each time point are reported in Table 1. Table 2 shows by subtest the count of boys in each group who received a raw score of 0, which was treated as missing as described previously. Table 3 provides F-tests for all of the fixed effects in the model. There is strong support for all model effects with the exception of the three-way interaction. The continuous variables (e.g., age) in the model are mean centered, so this effect represents differences at the mean of those variables. Tests are Benjamini-Hochberg adjusted for false discovery rate (FDR; Benjamini & Hochberg, 1995).

Between-Group Comparisons

The significant interaction of group with scale (see Table 3) addresses the question of between-group differences on the CASL subtests. The between-group differences, adjusted means, and standard errors for the three CASL subtests are presented in Table 4. On all three subtests (Antonyms, Syntax Construction, and Pragmatic Judgment), the TD group scored significantly higher (i.e., showed more skill) than all three other groups (p values < .05). Boys with FXS-O and FXS-ASD scored significantly higher than boys with DS in Syntax

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Construction. The FXS-O group scored significantly higher than both the DS and FXS-ASD groups on the Pragmatic Judgment subtest.

Results revealed strong group differences in Pragmatic Judgment scores after controlling for Antonyms and Syntax Construction scores in addition to nonverbal MA and maternal education. The TD group scored significantly higher than all other groups, as they did before adding Antonyms and Syntax Construction performance as covariates. The DS group did not differ significantly from either of the FXS groups. However, the boys with FXS-ASD scored significantly lower than did the boys with FXS-O.

Within-Group Comparisons

The diagnostic group X subtest interaction term in Table 3 addresses between-scale differences within each group. The pattern of results varied between groups. The TD boys scored significantly higher on the Antonyms subtest than on the Syntax Construction and Pragmatic Judgment subtests, with no significant differences between syntax and pragmatic performance. Significant differences were found between all three subtests for the FXS-O and DS groups, who showed similar patterns. Both groups scored highest on Antonyms, followed by Pragmatic Judgment, and finally Syntax Construction. Boys with FXS-ASD showed no significant difference between Pragmatic Judgment and Syntax Construction, but performance on both of these subtests was significantly below the Antonyms subtest.

Change over Time

The HLM also tested for group differences in the overall rate of change. The diagnostic group X chronological age interaction was significant, as shown in Table 3. As illustrated in Figure 1, the TD group showed a significantly steeper slope (.90, SE = .09) than FXS-O (.22, SE = .04), FXS-ASD (.22, SE = .04), and DS (.23, SE = .05) groups, with no other differences between the groups. The 3-way interaction in the model (see Table 3) further tested whether change over time differed between subtests within each group. This test was non-significant, suggesting that change over time within each group does not differ by subtest.

Autism Severity and Language in FXS

We replicated the original models but limited analysis to the FXS sample (combining FXS-O and FXS-ASD) and replaced diagnosis in the models with autism severity. In the model that included all three subtests, the significant interaction of severity with subtest, $F(2,401) = 7.10$, $p = 0.0009$, indicates that severity of autism symptoms does differentially impact the subtest scores. Only the slope for Pragmatic Judgment was significant (−1.71, SE = .73, $p = 0.0199$), with increasing severity related to lower Pragmatic Judgment scores, and this slope differed significantly from the slopes for Antonyms and Syntax Construction (p values < .01). The next model examined Pragmatic Judgment as the sole outcome, with Antonyms and Syntax Construction as covariates (parallel to the planned follow-up analysis described above). The effect of autism severity was again significant (−1.53, SE = .51, $p = .0035$), indicating that Pragmatic Judgment scores tended to decrease as autism severity increased.

Discussion

In this study, we compared three domains of language production over time within and across groups of boys with FXS with and without ASD, boys with DS, and younger TD boys of similar nonverbal MA. Although both FXS (Abbeduto et al., 2007, Roberts et al., 2008, Finestack et al., 2009) and DS (Chapman et al., 2002, Laws & Bishop, 2003, Martin et al., 2009) are known to be associated with language impairment, studies comparing expressive language domains longitudinally within and across groups have been lacking. In

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the present study, TD boys showed higher vocabulary, syntax, and pragmatic language skills than did boys with FXS-O, FXS-ASD, and DS; boys with FXS-O and FXS-ASD scored higher in syntax than boys with DS; and boys with FXS-O showed higher pragmatic skills than boys with FXS-ASD. Boys with FXS-O and boys with DS showed similar within-group patterns; both groups scored highest in vocabulary, followed by pragmatic language, and scored lowest in syntax. Boys with FXS-ASD also scored highest in vocabulary, but showed similar levels of pragmatic language and syntax ability. Overall the TD group showed significantly more change over time than all other groups.

These findings build on prior work in several important ways. Our finding that TD boys showed higher vocabulary skill than the other groups is consistent with several previous studies of boys with FXS (Roberts et al., 2007a, Sudhalter et al., 1991) and children with Down syndrome (Caselli et al., 2008, Chapman et al., 1998, Hick et al., 2005, Roberts et al., 2007c). For boys with FXS, previous findings had been mixed (Roberts et al., 2007c). These findings support prior reports that expressive vocabulary is indeed delayed relative to MA in FXS, but future research may utilize multiple assessments of vocabulary (e.g., antonym, synonym, and picture naming tasks; natural language sampling) in a single study for a more in-depth understanding of word knowledge abilities. TD boys in the current study also scored higher in syntax than did boys in all other groups, even after controlling differences in nonverbal MA and maternal education, and boys with FXS-O and FXS-ASD scored significantly higher than boys with DS in syntax. For males with FXS compared with TD males, these findings are consistent with those of recent studies (Estigarribia et al., 2010, Price et al., 2008, Roberts et al., 2007a) and contrary to some older reports (Ferrier et al., 1991, Sudhalter et al., 1991), again providing important evidence that expressive syntax is indeed delayed relative to MA in males with FXS. Our results are also consistent with the vast amount of literature supporting syntax delays in individuals with DS that are beyond expectations for cognitive level (e.g., Caselli et al., 2008, Chapman et al., 1998, Price et al., 2008) and with one additional study which found that boys with FXS produced more complex syntax than boys with DS during a natural language sample (Price et al., 2008).

For group comparisons of pragmatic language ability, we additionally controlled for differences in vocabulary and syntax skills, to ensure that any differences observed were not attributable to differences in base-line language ability (Tager-Flusberg, 2004). Results of the present study showed that TD boys scored significantly higher than all other groups in pragmatic language. For boys with FXS, this finding is not surprising given the ample evidence that males with FXS have impaired pragmatic language (Roberts et al., 2007b, Wolf-Schein et al., 1987, Sudhalter & Belser, 2001). For children with DS, the literature based on natural language samples and narratives suggests a complex pragmatic profile of strengths (Beeghly et al., 1990, Coggins et al., 1983) and weaknesses (Abbeduto et al., 2006, Abbeduto et al., 2008, Tannock, 1988, Roberts et al., 2007b). Results of the present study provide additional evidence that pragmatic language is delayed relative to MA expectations in boys with DS according to a standardized assessment. Finally, boys with FXS-ASD showed less pragmatic skill than did boys with FXS-O. Similarly, in complementary analysis using a continuous approach to ASD in FXS, increasing autism severity was related to lower pragmatic skills in the full FXS group. These findings add to the literature pointing to language differences among children with FXS based on autism status (Bailey et al., 2001, Philofsky et al., 2004, Rogers et al., 2001) and with regard to pragmatic language in particular (Roberts et al., 2007b).

Several significant within-group differences were also found, which interestingly was not the case for boys with FXS or DS in a previous study of receptive language across domains (Price et al., 2007) and underscores again the particular importance of expressive language in both groups. Both boys with FXS-O and boys with DS scored highest in vocabulary,

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followed by pragmatic language, and finally syntax. For boys with DS, this finding is consistent with previous evidence that expressive vocabulary is a relative strength compared with syntax (Laws & Bishop, 2003), and also extends this literature to suggest that pragmatic language, at least according to a standardized assessment, represents an intermediate challenge. The pattern of within-group findings was notably different for boys with FXS-ASD, who like the other groups performed highest in vocabulary, but unlike boys with FXS-O and DS did not show a relative strength in pragmatic language compared with syntax. This finding suggests not only differences between boys with FXS-O and FXS-ASD on an individual language domain, as previously discussed, but also differences in the strengths and weaknesses of these groups. This suggests that autism in the context of FXS affects language development differently than does ID or FXS alone. Finally, the TD boys showed a faster rate of growth overall than all other groups. Change over time within each group did not differ by subtest, suggesting that TD boys are developing expressive language skills more quickly across domains.

Study Strengths and Limitations

The present study has several strengths. First, we examined three domains of language production prospectively. Studies of language growth over time are of critical importance in understanding the various language strengths and weaknesses of different developmental disabilities, and how they may change over development. Second, sample sizes were large relative to many previous investigations, permitting us to interpret group profiles with relative confidence. Third, groups were similar on MA at the initial assessment (and FXS-O, FXS-ASD, and DS groups were not significantly different from each other at any time point). As expected, a disparity emerged over time on MA between the TD and other groups (the differences are significant at visits two and three), but we controlled for MA in analysis to determine whether expressive language profiles of boys with FXS or DS are reflective of developmental level or ID in general. Fourth, we controlled for maternal education and, in the case of pragmatic language, structural language ability, reducing the chance that observed group differences in language patterns stem from differences in socioeconomic status (SES), or that pragmatic language findings stem from differences in language functioning in general. Finally, to examine the role of autism in language in FXS, boys with FXS with and without ASD were included. Given increasing evidence that individuals with FXS and ASD may present with qualitatively different language-related impairments, it is important (clinically and theoretically) to characterize how the language development of this group may differ from children with FXS only.

This study also has some limitations, which may inform forthcoming investigations. First, there are clear receptive language demands of the Pragmatic Judgment and Syntax Construction subtests, but we did not include measures of receptive language in the analyses. Language comprehension may be a relative weakness in individuals with idiopathic autism (Hudry et al., 2010) and FXS-ASD (Lewis et al., 2006, Philofsky et al., 2004) and receptive vocabulary in particular may be related to autism severity in FXS (McDuffie et al., 2012). Thus, it is possible that the unique profile associated with FXS-ASD in the present study (i.e., relative weakness in pragmatic language) could reflect to some extent language comprehension differences between groups. Future studies should include measures of receptive language across domains to examine in relation to expressive language data. Second, we used three subtests from a single standardized assessment (the CASL), which may not be sufficient for characterizing differences in developmental trajectories across domains (Hoffman et al., 2011). Future investigations should utilize multiple assessments to more fully characterize development in the different domains of language in boys with FXS and DS. Third, autism group status was determined only with the ADOS (Lord et al., 2001). The ADOS is a direct assessment tool of current functioning with

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a trained evaluator in one situation. The other gold standard autism diagnostic measure, the Autism Diagnostic Interview-Revised (ADI-R; Lord et al., 1994), collects information from a parent about functioning over time in varying contexts and should additionally be used in future studies to ensure more valid autism classifications. Moreover, an idiopathic ASD group should be included in future studies to examine whether language profiles are similar or different in FXS and idiopathic cases of autism. Fourth, the TD group on average achieved a higher age equivalent score on the Antonyms subtest compared with the Syntax Construction and Pragmatic Judgment subtests. This finding was unexpected given that the CASL is a standardized measure, and may challenge the validity of the Antonyms test in this study. That said, between- and within-group findings for the Antonyms subtest are consistent with the existing literature on vocabulary skills for boys with FXS and DS, supporting its validity in the present investigation. Fifth, age equivalent scores have been shown to have some undesirable properties (see Mervis & Klein-Tasman, 2004). These scores tend to have larger standard errors than do standard scores, and their metric (months or years) does not represent true interval level information. In young developmentally delayed populations, however, standards scores often cannot be calculated due to floor effects (i.e., raw scores fall outside of the range where they can be converted to standard scores). Raw scores can be calculated but are metric-free, and do not provide values that can be readily interpreted. Age-equivalent scores, on the other hand, have the advantage of being in the metric of interest for the current research, namely change in our dependent variables over a range of age. Finally, the significant findings after controlling for nonverbal cognitive ability suggest of course that other factors beyond cognition are accounting for language development of children from these groups. However, with the exception of autism severity in the FXS group, we did not examine potential underlying mechanisms of language performance, such as anxiety, oral-motor skills and speech intelligibility, social cognition, and executive function, and whether relationships differ by group. Future studies should investigate predictors of individual differences to better understand between- and within-group differences and potentially affect language interventions.

Clinical Implications

Although assessment and intervention should be individualized to address the particular needs of a child and family, findings of the current study do have some important clinical implications. First, language production across the domains of vocabulary, syntax, and pragmatics should be addressed during assessment and intervention for boys with FXS and boys with DS. Second, interventions may build on relative strengths in vocabulary for all groups. Differences between the disability groups and within-group findings are suggestive of distinct language profiles, and support the need for tailored language interventions, perhaps focusing on different aspects of language for these different groups. For instance, expressive syntax may be a particularly significant target for boys with DS (who scored lower than all other groups), and also for boys with FXS, whereas pragmatic language would be an important focus of intervention for boys with both FXS and ASD. In sum, findings from this longitudinal study of language development in children with FXS (with and without ASD) and children with DS identified some important similarities and differences across these genetically-based syndromes and suggest several important areas for further investigation.

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What This Paper Adds

Both fragile X syndrome (FXS) and Down syndrome (DS) are associated with significant language delay, with language production more impaired than comprehension and some evidence that language impairments are more severe among individuals with both FXS and autism than FXS only. Studies comparing expressive language across domains over time and within groups are lacking, especially for boys with FXS, nor is it clear how autism in the context of FXS may influence language development across domains. In addition to providing evidence in areas where the extant literature is mixed, our longitudinal data demonstrate a slower rate of growth in expressive language across domains for boys with FXS and DS compared with typically developing controls. Our findings also suggest that boys with FXS without autism and boys with DS show a similar profile of strengths and weaknesses across language domains (scoring lowest in syntax) that differentiates them from boys with FXS with autism (who show similar levels of impairment in pragmatics and syntax) and may suggest differing areas of emphasis for language assessment and intervention.

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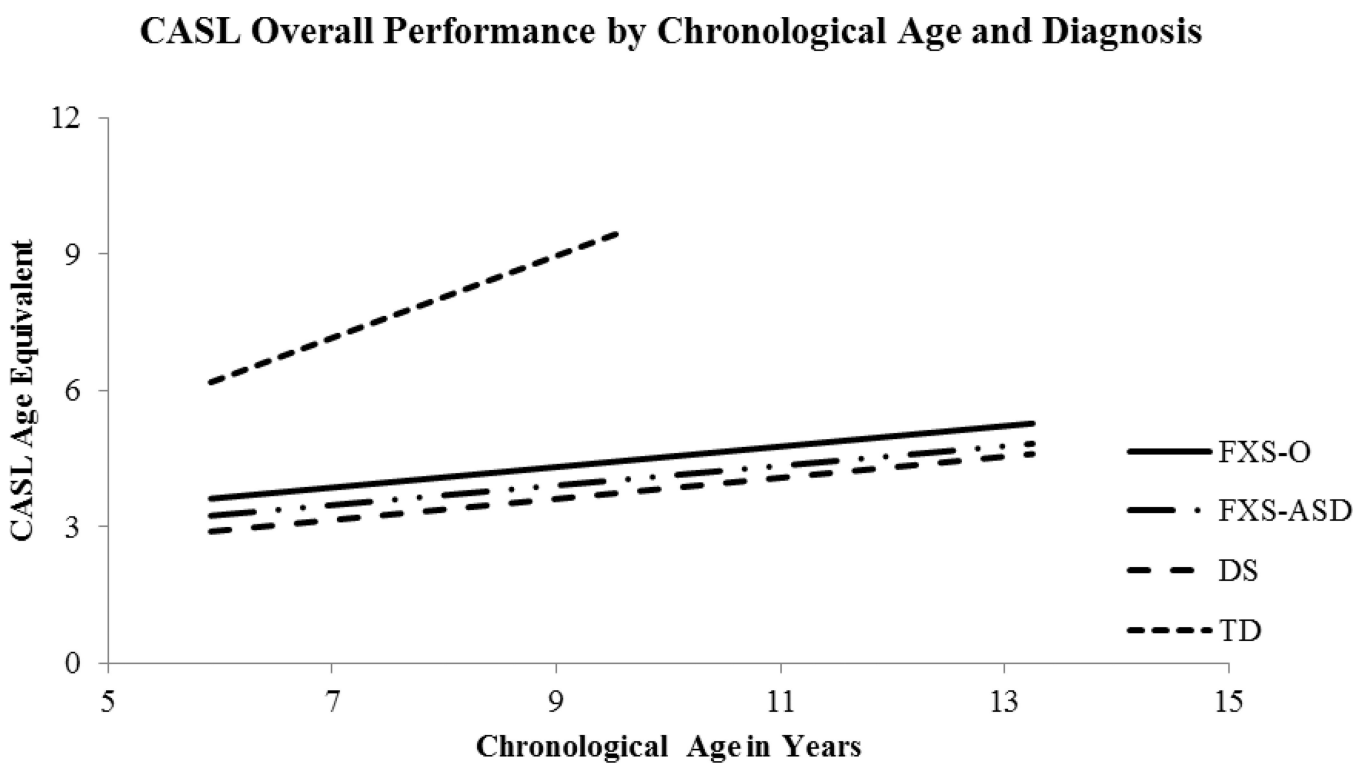

Figure 1.
Mean trajectories across scales within each group (representing a model-based mean). The TD group shows a significantly steeper slope than FXS-O, FXS-ASD, and DS groups, but change over time within each group does not differ by subscale. *Note.* CASL = Comprehensive Assessment of Spoken Language; FXS-O = fragile X syndrome only; FXS-ASD = fragile X syndrome with autism spectrum disorder; DS = Down syndrome; TD = typically developing.

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Table 1

CASL Age Equivalent Scores in Years and Background Variables

	FXS-O		FXS-ASD		DS		TD	
	N	**Mean (SD) Range**	**N**	**Mean (SD) Range**	**N**	**Mean (SD) Range**	**N**	**Mean (SD) Range**
Chronological age (in years)								
Visit 1	29	10.7 (3.1)	40	10.2 (3.1)	34	10.1 (2.9)	48	5.2 (1.1)
		3.9 – 14.5		4.4 – 15.5		5.4 – 16.0		3.1 – 8.2
Visit 2	26	11.5 (3.2)	32	11.5 (3.1)	21	10.4 (3.3)	36	6.5 (1.2)
		5.0 – 15.9		5.4 – 17.1		6.4 – 17.0		4.1 – 8.6
Visit 3	21	13.3 (2.7)	25	12.7 (3.0)	16	11.5 (3.3)	23	7.3 (1.1)
		6.0 – 16.9		6.5 – 17.8		7.4 – 17.9		5.5 – 9.5
Leiter-R mental age (in years)								
Visit 1	29	5.2 (0.8)	40	5.1 (0.7)	34	5.2 (1.0)	48	5.5 (1.4)
		3.6 – 6.6		2.9 – 6.5		2.8 – 8.2		3.6 – 11.2
Visit 2	26	5.3 (0.8)	32	5.3 (0.5)	21	5.5 (1.2)	36	7.2 (1.5)
		3.7 – 7.7		4.3 – 6.3		3.8 – 8.0		4.7 – 10.3
Visit 3	21	5.4 (0.7)	25	5.5 (0.5)	16	6.1 (1.4)	23	8.4 (1.8)
		3.9 – 7.7		4.7 – 6.7		4.0 – 9.0		5.3 – 12.4
Antonyms								
Visit 1	29	4.6 (1.4)	36	4.5 (1.6)	27	4.5 (1.7)	48	5.9 (1.7)
		2.8 – 7.7		2.8 – 8.7		2.8 – 8.7		2.9 – 9.7
Visit 2	25	5.3 (1.3)	30	5.2 (1.6)	19	5.4 (1.8)	35	7.3 (2.1)
		2.8 – 7.7		3.1 – 9.7		2.9 – 8.7		3.9 – 12.5
Visit 3	10	5.2 (0.9)	13	5.7 (1.5)	5	5.0 (0.8)	12	9.5 (2.5)
		3.9 – 6.5		3.1 – 8.0		4.2 – 6.2		5.9 – 13.8
Syntax Construction								
Visit 1	29	3.7 (1.3)	40	3.5 (1.4)	32	3.1 (1.5)	48	5.2 (1.6)
		2.1 – 6.3		2.1 – 6.3		2.1 – 6.8		2.6 – 10.3
Visit 2	26	3.8 (1.4)	32	4.0 (1.6)	21	3.5 (1.6)	36	7.0 (1.8)
		2.2 – 7.1		2.1 – 7.8		2.1 – 7.5		4.0 – 11.0
Visit 3	21	4.3 (1.8)	25	4.3 (1.5)	16	4.1 (2.0)	23	8.1 (2.3)

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		FXS-O		FXS-ASD		DS		TD	
		N	**Mean (SD) Range**	**N**	**Mean (SD) Range**	**N**	**Mean (SD) Range**	**N**	**Mean (SD) Range**
			2.2 – 9.0		2.1 – 6.8		2.2 – 7.8		5.2 – 14.3
Pragmatic Judgment									
	Visit 1	28	4.5 (1.7)	37	3.8 (1.1)	32	3.8 (1.4)	48	5.6 (1.6)
			2.4 – 9.2		2.4 – 6.3		2.4 – 7.7		2.4 – 9.8
	Visit 2	25	4.5 (1.4)	32	4.3 (1.3)	20	4.1 (1.1)	36	7.2 (1.6)
			2.8 – 8.5		2.6 – 7.3		2.6 – 6.5		3.4 – 11.2
	Visit 3	21	5.1 (1.6)	25	4.5 (1.3)	15	5.2 (1.6)	23	8.3 (2.0)
			3.0 – 9.3		2.6 – 7.3		2.8 – 8.8		4.9 – 12.8
Maternal education (visit 1, in years)		29	13.7 (2.1)	40	15.2 (2.2)	34	16.1 (2.2)	48	16.2 (2.2)
			12.0 – 20.0		12.0 – 20.0		12.0 – 20.0		12.0 – 20.0
Race/Ethnicity		N	%	N	%	N	%	N	%
Caucasian		25	86.2	35	87.5	29	85.3	34	70.8
African American		4	13.8	3	7.5	5	14.7	9	18.8
Other		0	0	2	5.0	0	0	5	10.4

Note. CASL = Comprehensive Assessment of Spoken Language; FXS-O = fragile X syndrome only; FXS-ASD = fragile X syndrome with autism spectrum disorder; DS = Down syndrome; TD = typically developing; Leiter-R = Leiter International Performance Scale-Revised.

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Table 2

Number (percent) of Observations[1] and Boys by Group and CASL Subtest Receiving a Raw Score of 0

	FXS-O	FXS-ASD	DS	TD
Antonyms				
Observations	2 (3%)	5 (6%)	9 (15%)	0
Boys	2 (7%)	4 (10%)	6 (18%)	0
Syntax Construction				
Observations	0	0	1 (1%)	0
Boys	0	0	1 (3%)	0
Pragmatic Judgment				
Observations	2 (3%)	3 (3%)	4 (6%)	0
Boys	2 (7%)	3 (8%)	2 (6%)	0

Note. CASL = Comprehensive Assessment of Spoken Language; FXS-O = fragile X syndrome only; FXS-ASD = fragile X syndrome with autism spectrum disorder; DS = Down syndrome; TD = typically developing.

[1] All observations for a given subtest across time points, where the same boy may receive a raw score of 0 more than once.

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Table 3

Tests of Individual Effects for CASL Analysis

Effect	Test
Mean effects	
Diagnostic group	$F(3,805) = 53.92^{***}$
Subtest	$F(2,805) = 61.28^{***}$
Diagnostic group × subtest	$F(6,805) = 3.71^{***}$
Slope effects	
Chronological age (CA)	$F(1,805) = 136.42^{***}$
CA × subtest	$F(2,805) = 6.66^{*}$
Diagnostic group × CA	$F(3,805) = 20.49^{***}$
Diagnostic group × CA × subtest	$F(6,805) = 0.77$
Covariates	
Leiter-R mental age	$F(1,805) = 30.05^{***}$
Maternal education	$F(1,805) = 21.56^{***}$

Note. CASL = Comprehensive Assessment of Spoken Language; Leiter-R = Leiter International Performance Scale-Revised.

$^{*}p < .05$;

$^{**}p < .01$;

$^{***}p < .001$.

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Table 4

Adjusted Means (adjusted for Leiter-R mental age and maternal education), Standard Errors, and Between-Group Differences by CASL Subtest

	FXS-O	FXS-ASD	DS	TD
Antonyms	4.91 (0.22)[a]	4.53 (0.19)[a]	4.46 (0.20)[a]	9.91 (0.43)[b]
Syntax Construction	3.84 (0.22)[a]	3.67 (0.18)[a]	3.01 (0.20)[b]	9.26 (0.42)[c]
Pragmatic Judgment	4.58 (0.23)[a]	3.90 (0.18)[b]	3.76 (0.20)[b]	9.19 (0.42)[c]

Note. CASL = Comprehensive Assessment of Spoken Language; Leiter-R = Leiter International Performance Scale-Revised; FXS-O = fragile X syndrome only; FXS-ASD = fragile X syndrome with autism spectrum disorder; DS = Down syndrome; TD = typically developing. Different superscripts within a row indicate significant differences. If groups share the same letter, differences were not significant.