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Abstract

Introduction: Although reading skills are critical for the success of individuals with intellectual disabilities, literacy has received little attention in fragile X syndrome (FXS). This study examined the literacy profile of FXS. *Methods:* Boys with FXS ($n = 51$; mean age 10.2 years) and mental-age-matched boys with typical development ($n = 35$) participated in standardized assessments of reading and phonological skills. *Results:* Phonological skills were impaired in FXS, while reading was on-par with that of controls. Phonological awareness predicted reading ability and ASD severity predicted poorer phonological abilities in FXS. *Conclusion:* Boys with FXS are capable of attaining reading skills that are commensurate with developmental level and phonological awareness skills may play a critical role in reading achievement in FXS.

Keywords: fragile X syndrome; autism spectrum disorder; literacy; reading; phonological skills; phonological awareness

Reading and Phonological Skills in Boys with Fragile X Syndrome

Recent research in literacy and developmental disabilities has challenged the assumption that children with intellectual disabilities cannot attain functional levels of reading (Boudreau, 2002; Conners, Rosenquist, Sligh, Atwell, & Kiser, 2005; Cossu, Rossini, & Marshall, 1993; Laing, 2002; Levy, Smith, & Tager-Flusberg, 2003). However, it is clear that significant variability exists in the level of reading achievement of children with intellectual disabilities (Boudreau, 2002; Conners, 2003; Katims, 1994, 1996, 2000). Functional reading skills are critical for the success of individuals with intellectual disabilities, as reading achievement is associated with increased vocational opportunities, increased peer acceptance, and greater independence in daily living activities (Erickson, 2000; Miller, Leddy, & Leaveitt, 1999). Despite the importance of reading skills for supporting autonomy in individuals with intellectual disabilities, literacy has received little attention in fragile X syndrome, the most common inherited cause of intellectual impairment.

Fragile X Syndrome

FXS is a single gene, X-linked neurodevelopmental disorder with an estimated prevalence of 1 in 4,000 males and 1 in 8,000 females (Sherman, Pletcher, & Driscoll, 2005; Turner, Webb, Wake, & Robinson, 1996). FXS is caused by an expansion of the cytosine-guanine-guanine trinucleotide repeat on the *Fragile X Mental Retardation-1* (*FMR1*) gene located on the long arm of the X chromosome. In full mutation FXS, the *FMR1* gene methylates, or "shuts down", and impedes the normal production of FMRP, an important protein for brain development. Because the syndrome is X-linked, males are typically more impaired than females who have a second, functional X chromosome. Most males with FXS have moderate intellectual impairment, although cognitive ability is unevenly affected and some males demonstrate skills in the low-average range (Abbeduto & Hagerman, 1997; Hagerman, 2002; Hagerman et al., 1994). The cognitive domains of working memory (Baker et al., 2011; Lanfranchi, Cornoldi, Drigo, & Vianello, 2008; Ornstein et al., 2008), sequential processing (Burack et al., 1999; Dykens, Hodapp, & Leckman, 1987), and attention (Cornish, Scerif, & Karmiloff-Smith, 2007;

Mazzocco, Pennington, & Hagerman, 1993; Ornstein et al., 2008; Sullivan et al., 2006) are particularly

affected, with deficits in these areas greater than mental age-based expectations.

Almost all individuals with FXS exhibit the characteristic features of autism spectrum disorders

(ASD), such as poor eye contact, hand flapping, hand biting, perseveration, and social and

communication deficits (Hagerman et al., 1986; Merenstein et al., 1996; Reiss & Freund, 1992). Sixty to

seventy-four percent of boys with FXS exhibit sufficient behaviors to meet diagnostic criteria for ASD

(Klusek et al., 2014; Garcia-Nonell, 2008). ASD comorbidity in FXS has a detrimental impact on

developmental outcomes, including language outcomes such as receptive (Lewis et al., 2006; Rogers,

Wehner, & Hagerman, 2001), expressive (Philofsky, Hepburn, Hayes, Hagerman, & Rogers, 2004), and

pragmatic language abilities (Klusek, Martin, & Losh, 2014; Losh, Martin, Klusek, Hogan-Brown, &

Sideris, 2012; Roberts, Martin, et al., 2007). Although the impact of ASD on the literacy skills of

individuals with FXS is unknown, poor language skills are a risk factor for reading difficulties (Bishop &

Snowling, 2004; Catts & Kamhi, 2005; Nation, Clarke, Marshall, & Durand, 2004), and thus it would be

expected that ASD increases the risk for literacy failure in children with FXS.

Literacy Skills of Individuals with Fragile X Syndrome

Few studies have investigated reading abilities of individuals with FXS, and little is known about

processes that may support literacy in this population, such as phonological skills. However, it is clear

that literacy skills are significantly impaired in FXS. In a national survey of 1,105 families of children

with FXS, Bailey et al. (2009) found that only 19% of adult males with FXS were reported to read books

containing new words or concepts. Significant impairments were also detected in basic literacy skills, as

only 44% of adult males with FXS were reported to read basic picture books and 59% were reported to

know letter sounds. Furthermore, although boys with FXS gained literacy skills from birth through age

five, Bailey et al. (2009) detected a developmental plateau in the acquisition of literacy skills at around

six to ten years. Similar developmental plateaus, occurring at approximately ten years of age, have been

reported for the acquisition of letter/word recognition skills (Roberts et al., 2005) and phonological

awareness skills (Adlof et al., 2014) in boys with FXS, suggesting literacy acquisition in FXS slows in

late childhood. While reading in FXS may be delayed relative to age-based expectations, a preliminary study by Johnson-Glenberg (2008) found that word identification skills of males with FXS (*n* = 13) were *better* than those of younger, typically developing boys who were matched on nonverbal mental age. Thus, basic reading skills may be a strength for males with FXS relative to general cognitive ability.

In contrast, phonological skills (i.e., skills in "the domain of language that pertains to the elements of speech and the systems that govern structural relationship among these elements within and across words;" Scarborough & Brady, 2002, p. 303) appear to lag behind cognitive expectations. Phonological ability encompasses a broad range of skills such as the encoding of phonological information in short-term memory, the use of phonological codes in working memory, the retrieval of phonological labels from long-term memory, and the manipulation of the phonological structure of words (Adams, 1990; Catts & Kamhi, 1999; Wagner & Torgesen, 1987; Whitehurst & Lonigan, 2002). In particular, phonological awareness, or "attending to, thinking about, and intentionally manipulating the phonological aspects of spoken language" (Scarborough & Brady, 2002, p. 312), is a skill set highly predictive of reading ability in typical development (Bird, Bishop, & Freeman, 1995; Ehri & Wilce, 1985; Liberman, Shankweiler, Fischer, & Carter, 1974; Perfetti, Beck, Bell, & Hughes, 1987; Share & Stanovich, 1995). In a longitudinal investigation of phonological awareness in 54 school-aged boys with FXS, Adlof and colleagues (2014) found that boys with FXS exhibit lower level phonological awareness skills than younger, mental-age matched typically developing boys. However, no group differences were detected in the rate of change over time, indicating that phonological awareness growth in FXS is commensurate with cognitive development.

Phonological skills in FXS may also be weaker than would be expected based on basic reading abilities. Johnson-Glenberg (2008) found that boys with FXS (*n* = 13) who were matched with typically developing children on word identification skills showed phonological decoding skills (measured by performance on a word attack task) that lagged behind those of the controls by about two years. Clinical reports by Braden (2002) and Spiridigliozzi, et al. (1994) have also supported weaknesses in word decoding skills and relative strengths in familiar word decoding in FXS. Given these reports, it has been

suggested that children with FXS rely on different sub-processes to identify words than do typically developing children, with greater dependence on a gestalt or "whole-word" approach to word decoding. Yet, emerging evidence suggests that, despite relative weaknesses in this domain, phonological ability may be an important predictor of reading achievement in FXS. In one study of 54 boys with FXS, phonological awareness accounted for significant variability in both concurrent and later letter/word identification skills (Adlof et al., 2014).

Potential impact of ASD on literacy in FXS. As previously discussed, individuals with FXS are at elevated risk for ASD and relatively little is known about the impact of ASD symptoms on literacy skills in FXS, despite a documented impact of ASD symptoms on language outcomes in this population. Research in this domain is limited to a single report by Adlof and colleagues (2014), which did find evidence that ASD symptoms influenced the level or rate of phonological awareness growth in a sample of 54 school-aged boys with FXS. ASD symptoms have not yet been examined as potential contributors to reading or other phonological deficits in FXS. In idiopathic ASD, there is a significant variability in literacy level that is consistent with the substantial clinical heterogeneity seen in the disorder (Nation et. al., 2006). Among high-functioning individuals, basic reading skills generally fall within the average range, whereas reading comprehension is often impaired (Huerner & Mann, 2010; Nation et al., 2006; Jones et al., 2009). However, not all individuals have basic reading skills that are within normal limits; Nation et al. (2006) examined children with ASD of all functioning levels and found that about 20% were completely unable to read. Thus, literacy is impaired in idiopathic ASD and it is possible that ASD symptoms may negatively impact literacy achievement in FXS.

The Present Study

While existing publications shed some light on the literacy profile of individuals with FXS, research is needed to more fully investigate patterns of strengths and weaknesses, areas of atypical development, and predictors of literacy achievement in this population. This study addressed the following questions:

1. What is the level of reading and phonological ability in school-aged boys with FXS?

2. Do reading and phonological skills of boys with FXS differ from those of typically developing boys matched on nonverbal cognitive ability?

3. What are strengths and weaknesses of the reading and phonological profiles among boys with FXS?

4. Do phonological awareness skills (i.e., explicit knowledge of individual phonemes within words) predict reading ability in boys with FXS?

5. What is the impact of ASD symptoms on the reading and phonological skills of boys with FXS?

Methods

Participants

Fifty-one boys with full mutation fragile X syndrome (FXS) and a control group of 35 mental age-matched boys with typical development (TD) were drawn from a larger longitudinal study of development and achievement in children with FXS. Participants with FXS were recruited through genetics clinics, developmental evaluation centers, and early intervention programs in the Southeastern United States. The diagnosis of full mutation FXS was confirmed by DNA report. Participants with TD were recruited from childcare centers, preschools, and local elementary schools.

The larger study employed a group matching procedure, where participants meeting study criteria were enrolled until pre-determined recruitment goals were achieved. The mean nonverbal mental ages of the groups were examined periodically during ongoing recruitment and targets were honed as necessary to prioritize enrollment of participants who best facilitated group-level matching, based on caregiver report during initial phone screening. This strategy was successful and resulted in groups well matched on nonverbal mental age, according to the criteria outlined by Mervis & Robinson (2003) with a p-value >.50. All FXS and TD participants with available literacy data were included in the present study and these groups did not differ on nonverbal mental age ($p = .628$) as measured with the Brief IQ Scale of the Leiter International Performance Scale-Revised (Leiter-R; Roid & Miller, 1997). The mean age of the boys with FXS was 10.2 years (range 7.9-13.2) and 5.1 years (range 3.3-7.4) for the boys with TD; the groups differed significantly in chronological age ($p = .001$). The maternal education level of the groups

also differed; this variable was controlled for in analyses. Participant characteristics are presented in Table 1. Phonological awareness data from a subset of the participants in this study have been previously reported in a longitudinal study of phonological awareness in XX et al. (2014).

[Table 1 top]

Procedures

Procedures were approved by the Institutional Review Board of the XX. Assessments were administered as part of a broader protocol by trained research associates (most of whom were either doctoral or master's level clinicians). To increase compliance, assessments took place at the child's school during regular school hours.

Measures

Nonverbal cognition. The Brief IQ composite of the Leiter International Performance Scale-Revised (Leiter-R; Roid & Miller, 1997) was used to measure nonverbal cognitive ability. The present study sought to elucidate how children with FXS differed from children of a similar developmental level on measures of reading and phonology. The Leiter-R, a nonverbal cognitive measure, was chosen as the most optimal index of intellectual functioning for the present analyses, given the potential for language-based assessment to create bias or reduce accuracy in the cognitive appraisal of populations with known language impairments, such as FXS (i.e., Hooper et al., 2000). The Leiter-R Brief IQ is comprised of four subscales: Figure Ground, Form Completion, Sequential Order, and Repeated Patterns. The psychometric properties of the Leiter-R Brief IQ are excellent, with Cronbach's alphas ranging from 0.75-0.88 across subtests and support for concurrent validity (Tsatsanis et al., 2003; Hooper & Bell, 2006). The Leiter-R has been used extensively to characterize cognitive ability in populations with intellectual disabilities, including FXS (e.g., Glenn & Cunningham, 2005; Hooper et al., 2000; Skinner et al., 2005). Age equivalent scores were used in analysis.

Reading. Three subtests of the Woodcock-Johnson Tests of Academic Achievement-Revised (WJ-R; Woodcock & Johnson, 1990) were used to measure reading skills: Letter-Word Identification, Passage Comprehension, and Word Attack. Scores from Letter-Word Identification and Passage

Comprehension subtests were combined to form a Broad Reading composite score, the primary dependent variable examined. The Letter-Word Identification subtest measures symbol recognition, letter naming, and word naming. The Passage Comprehension subtest measures reading comprehension and vocabulary skills. Initial items require children to point to one picture from a choice of four represented by a phrase, while more difficult items measure their ability to provide a missing word in a passage. The Word Attack subtest measures decoding skills by requiring children to use knowledge of grapheme-phoneme correspondence to correctly decode unfamiliar (nonsense) words such as "jop" or "shamble". Although word attack tasks involve phonological decoding skills, this subtest was considered a "reading" task in this paper as it measures the ability to decode novel words (based on the assumption that decoding nonsense words requires the same cognitive processes as decoding a real novel word). Standard scores were used in analyses describing the performance of the FXS group compared to age-based expectations; W scores were used in group comparisons and predictive analyses (see *Data Analysis*). W scores are computed through a mathematical transformation of raw scores into Rasch-model scores, providing a mechanism to represent both the child's degree of mastery as well as item difficulty. The scores are centered at 500 for a ten year-old child. Because each possible raw score of the test is associated with a unique W score, the use of W scores may help resist floor effects by allowing for greater variability at the lower tail of performance to be captured than would standard scores. W scores also possess favorable psychometric properties in that they are norm-referenced (unlike raw scores) and on an equal-interval scale (unlike age equivalent scores); see Jaffe (2009).

Phonological skills. Four subtests from the Woodcock Johnson Tests of Cognitive Abilities-Third Edition (WJ-III; Woodcock et al., 2001) were used to index phonological ability: Sound Blending, Incomplete Words, Memory for Words, and Rapid Picture Naming. The combined scores from the Sound Blending and Incomplete Word subtests were used to calculate the Phonemic Awareness composite score, which was used as an index of overall phonological awareness skill. The Sound Blending subtest assesses skills in synthesizing sounds. For example, the participant is given the sounds "/k/ /a/ /t/?" and asked what word they make when put together. The Incomplete Words subtest involves naming a

complete word when given the word with missing phonemes, such as "What word am I trying to say? Alli_a_or?". The Memory for Words subtest measures short-term phonological memory by requiring the participant to repeat an increasingly longer series of syntactically and semantically unrelated words. The Rapid Picture Naming subtest assesses phonological processing skills through the naming of pictures of objects as quickly and accurately as possible. Standard scores and W scores were used in analyses (see *Data Analysis*).

ASD symptoms. The Childhood Autism Rating Scale (CARS; Schopler et al., 1988) assessed behavioral symptoms consistent with ASD. The CARS consists of 15 items that tap specific behaviors characteristic of ASD (e.g., relating to people, adaptation to change, verbal and nonverbal communication). Each item is rated on a scale from 1-4 and total scores above 30 are consistent with a diagnosis of ASD. Ten of the 51 boys with FXS (20%) scored above the cut-off for ASD. Prior research supports the utility of the CARS for capturing a continuum of autistic behaviors within individuals with a variety of developmental disorders, including FXS (e.g., Rellini, Tortolani, Trillo, Carbone, & Montecchi, 2004; Hatton et al., 2006; Sloneem, Oliver, Udwin, & Woodcock, 2011). Consistent with this work, this study used the CARS total score as a continuous index of autism symptom severity. Ratings were completed by consensus of two examiners immediately following the assessment. CARS data were not collected for the boys with TD, owing to the expected lack of ASD symptoms.

Data Analysis

Descriptive statistics were computed to describe the performance of the boys with FXS on the standardized assessments in comparison to published norms, using standardized scores. Then, Multivariate Analysis of Covariance (MANCOVA) was used to test group differences on the mean W scores for the reading and phonological subtests, controlling for mental age and maternal education level (indexed as a continuous variable representing the number of years of formal education). Potential false discovery rate was controlled for at the model level with the Benjamini-Hochberg method (Benjamini & Hochberg, 1995). Effect sizes (Cohen's *d*) were computed; generally, an effect size of "0.2" is considered small, "0.5" medium, and effects of "0.8" or greater are considered to be large (Cohen, 1988). Next,

patterns of strengths and weaknesses in reading and phonological skills in the FXS group were tested with

a series of dependent-samples *t*-tests, using W scores. Multiple comparisons were controlled using the

Benjamini-Hochberg procedure (Benjamini & Hochberg, 1995). Then, a series of multiple regression

models were used to examine phonological awareness (indexed by the Phonemic Awareness composite of

the WJ-III) as a predictor of readings skills (i.e., Letter-Word Identification, Passage Comprehension,

Word Attack, and the Broad Reading composite of the WJ-R) in FXS, after controlling for mental age and

maternal education level. Finally, ASD symptoms were examined as a predictor of literacy skills in the

group with FXS, controlling for mental age and maternal education level. For each regression model,

multicollinearity among predictors was evaluated via tolerance statistics, with no indication of problems

related to multicollinearity.

Results

Literacy Skills in FXS Relative to Chronological Age-Based Norms

Reading skills. Means, standard deviations, and ranges for the reading measures are presented in

Table 2. Reading skills were substantially delayed for the majority of the boys with FXS; 92% of the boys

obtained a Broad Reading standard score that was two standard deviations or more below the mean, with

an average Broad Reading score of 41.1 (*SD* = 19.0). On the Letter-Word Identification subtest, the

average standard score was 45.9 (*SD* = 18.6). Letter-Word Identification scores ranged from 3-84, with

94% scoring two standard deviations or more below the mean. On the Passage Comprehension subtest,

the average standard score was 44.0 (*SD* = 19.1). Ninety-two percent of the boys scored two or more

standard deviations below the mean on this subtest. Finally, 98% (49 of 51) of the boys had Word Attack

scores that were two or more standard deviations below the mean, with the group average at 57.6 (*SD* =

13.0). The percentage of boys with FXS scoring within normal limits (i.e., greater than or equal to 85,

within one standard deviation of the mean) on the reading subtests were as follows: 2% for Broad

Reading, 0% for Letter-Word Identification, 2% for Passage Comprehension, and 2% for Word Attack.

[Table 2 top]

Phonological skills. A wide range of phonological skills (see Table 3) was evident, with standard scores on the Phonemic Awareness composite ranging from 5-117. Forty-four percent of the boys exhibited significant delays in phonological awareness as evidenced by a standard score greater than or equal to two standard deviations below the mean on this composite. Forty-seven percent scored two standard deviations below the mean on the Sound Blending subtest, 30% for Incomplete Words, 98% for Memory for Words, and 55% for Rapid Picture Naming. The percentage of boys with FXS scoring within normal limits on the phonological subtests of the WJ-III are as follows: 22% for Sound Blending, 49% for Incomplete Words, 0% for Memory for Words, 20% for Rapid Picture Naming, and 27% for the Phonemic Awareness composite.

[Table 3 top]

Literacy Skills in FXS Relative to Mental Age-Matched Typically Developing Children

Reading skills. MANCOVA indicated a significant group effect on the WJ-R reading scores after covarying for mental age and maternal education (Pillai's Trace = 0.90, F [3, 76] = 215.05, $p <$ .001). Univariate analysis on individual subtests showed a significant group effect for the Broad Reading composite (F [1, 82] = 10.95, $p < .001$), with the boys with FXS exhibiting more advanced reading skills than the boys with TD ($d = .77$). The boys with FXS also showed stronger performance on the Passage Comprehension subtest (F [1, 82] = 12.98, $p = .001$; $d = .84$) and the Letter-Word Identification subtest (F [1, 82] = 4.55, $p = .036$; $d = .50$). Word Attack performance did not differ across the groups ($p = .515$, $d =.25$). Group comparisons and effect sizes are presented in Figure 1.

[Figure 1 top]

Phonological skills. MANCOVA indicated a significant group effect on the WJ-III phonological subtests (Pillai's Trace = 0.63, F [7, 71] = 24.48, $p < .001$). Univariate analysis of individual subtests showed significant group effects for the Phonemic Awareness composite (F [1, 75] = 7.13, $p = .009$, $d =$ .63) as well as for Sound Blending (F [1, 75] = 9.98, $p = .002$, $d = .75$) and Memory for Words (F [1, 75] = 101.24, $p < .001$, $d =2.39$), with lower skills in the group with FXS relative to TD. The groups did not

differ on Incomplete Words ($p = .140$, $d =.35$) or Rapid Picture Naming ($p=.836$, $d =.05$) performance.

Figure 2 presents groups comparisons and effect sizes.

[Figure 2 top]

Literacy Strengths and Weaknesses in FXS

Reading skills. Performance across the reading subtests differed significantly in the boys with

FXS, with the strongest performance on Word Attack, followed by Passage Comprehension, and then

Letter-Word Identification ($ps < .002$).

Phonological skills. Performance on the Incomplete Words subtest was a relative strength, with

significantly higher performance on this subtest than each of the other phonological subtests of the WJ-III

($ps < .001$). In contrast, short-term phonological memory was a relative weakness, with significantly

poorer performance on the Memory for Words subtest than the other subtests ($ps < .001$). Significant

differences were detected in performance on each of the phonological subtests, with the highest

performance in Incomplete Words, followed by Sound Blending, Rapid Picture Naming, and Memory for

Words ($ps < .032$).

Phonological Awareness as a Predictor of Reading Ability in Boys with FXS

In the group with FXS, phonological awareness accounted for 14% of the variance in Broad

Reading skills above and beyond the effects of mental age and maternal education (ΔF [1, 42] = 12.05, p

= .001). Phonological awareness also accounted for significant variance in the component Broad Reading

subtests: Letter Word Identification (ΔF [1, 2] = 9.13, p = .004; ΔR^2 = .14) and Passage Comprehension

(ΔF [1, 42] = 9.29, p = .004; ΔR^2 = .11). Phonological awareness did not account for unique variance in

Word Attack skills (ΔF [1, 40] = 0.93, p = .341).

ASD Symptoms as a Predictor of Literacy in Boys with FXS

ASD symptoms did not account for additional variance in reading skills on the Broad Reading (p

= .254), Passage Comprehension (p = .761), Letter-Word Identification (p = .106), or Word Attack (p =

.485) subtests of the WJ-R beyond the effects of mental age and maternal education. For the

phonological subtests of the WJ-III, elevated ASD symptoms predicted poorer performance on Sound

Blending (ΔF [1,44] = 4.59, p = .038; ΔR^2 = .06), Incomplete Words (ΔF [1,44] = 4.469, p = .040; ΔR^2 = .06), and Rapid Picture Naming (ΔF [1,43] = 7.29, p = .010; ΔR^2 = .09), after controlling for mental age. A trend was detected for ASD symptoms as a predictor of performance on the Phonemic Awareness composite (ΔF [1,42] = 3.03, p = .0894; ΔR^2 = .05). ASD symptoms were not a significant predictor of performance on the Memory for Words subtest (p = .116).

Discussion

This study aimed to (a) describe the level of literacy achievement and profiles of strengths and weaknesses among school-aged boys with FXS, (b) determine whether boys with FXS exhibit impaired literacy skills relative to younger, mental-age matched boys with TD, (c) determine whether phonological awareness skills predict reading ability in FXS, and (d) determine the impact of ASD symptoms on the literacy skills of boys with FXS. Although the reading skills of the boys with FXS were substantially delayed relative to chronological-age expectations, comparison with younger, mental age-matched boys with TD suggested that reading ability in FXS is on par with cognitive expectations (and on some subdomains, such as passage comprehension and letter-word identification, performance is more advanced than would be expected). In contrast, the boys with FXS exhibited phonological skills that were weaker than would be expected given cognitive ability. Despite apparent weaknesses in phonological ability, skills within this domain emerged as an important predictor of reading success; phonological awareness accounted for ~15% of variance in reading skills, above and beyond the effects of mental age and maternal education level. Overall, ASD symptoms did not impact the reading abilities of the boys with FXS, whereas a number of phonological skills were influenced by ASD symptoms.

Literacy Level in Boys with FXS

Considerable within-syndrome variation was detected, with standard scores ranging from average to clinically impaired. While a small percentage of boys had reading skills within normal limits (~2%), the vast majority of boys with FXS (i.e., 92-98%) exhibited significant delays. The finding of impaired reading performance on standardized tests corroborates evidence from caregiver-report indicating that the majority of males with FXS could not read books containing novel words or concepts (Bailey et al.,

2009). While few boys with FXS performed within normal limits on the reading subtests of the WJ-R,

about a fourth of the boys scored within one standard deviation of the mean on the phonological subtests

of the WJ-III, with considerable variability across individual subtests. The standard scores of the

relatively large sample in this study show that boys with FXS are not only capable of acquiring

phonological skills, but a minority have phonological abilities that are on par with age-based expectations

(0-49% across phonological subtests).

Profile of Literacy Strengths and Weaknesses

Short-term phonological working memory (measured with the Memory for Words subtest)

emerged as a weakness relative to all other phonological skills tested. Performance on this subtest was

significantly impaired (i.e., two standard deviations below the mean) for nearly all of the boys with FXS,

and none scored within normal limits on this measure. Working memory impairments are well

documented in FXS (Baker et al., 2011; Hooper et al., 2008; Ornstein et al., 2008), with some evidence

suggesting disproportionately affected phonological working memory relative to other domains, such as

visuospatial working memory (Pierpont, Richmond, Abbeduto, Kover, & Brown, 2011). Auditory

working memory has been found to contribute to poor language functioning in children with other

neurodevelopmental disabilities (Conners, Rosenquist, & Taylor, 2001) and may play a role in literacy

attainment as well. In contrast, phonetic processing on the Incomplete Words subtest was a strength

relative to other phonological skills, with nearly half of the boys with FXS scoring within one standard

deviation of the mean. This subtest indexes the ability to extract linguistic features such as the placement

and manner of articulation of consonants. While very few prior studies have reported on the processing of

phonetic information in FXS, others have found the production of phonological sounds and patterns to be

on par with cognitive expectations in boys with FXS (Barnes et al., 2009; Barnes, Roberts, Mirrett,

Sideris, & Misenheimer, 2006).

Given prior reports suggesting weaknesses in phonological decoding relative to whole-word

decoding (e.g., Braden 2002; Johnson-Glenberg, 2008; Spiridigliozzi, et al., 1994), it is unexpected that

Work Attack was a relative reading strength for the boys with FXS. However, this finding should be

interpreted with caution as the Word Attack scores across both groups represented floor-level performance on that subtest. Although it is possible to compare performance across WJ-R subtests because they are centered at approximately the same time point, the range of possible scores differs across the subtests; Word Attack is a later-emerging skill and thus W scores for this subtest do not extend as low as those of the other reading subtests examined. Because the intent of this study was to characterize literacy skills in boys with FXS as a population, we felt that it was important to include all participants, even those who did not obtain a basal on some subtests.

Group Comparisons

Although significant delays were detected relative to age-based norms, comparison with a mental age-matched sample of younger children with TD indicated that the reading levels of boys with FXS were *on par with* or *better than* mental age-based expectations. The boys with FXS outperformed the boys with TD on Passage Comprehension and the Broad Reading composite, and did not differ from the boys with TD on Letter-Word Identification. Phonological word decoding (i.e., word attack performance) in FXS was commensurate with cognitive expectations, which contrasts with prior clinical and preliminary reports that have suggested phonological word decoding weaknesses in this population (e.g., Braden 2002; Johnson-Glenberg, 2008; Spiridigliozzi, et al., 1994). These findings show that individuals with FXS are capable of achieving reading skills at or beyond cognitive expectations, diminishing the importance of IQ in any consideration of their underlying phonological processes and overall reading capabilities. The higher scores obtained by the boys with FXS may be related to their older chronological ages and longer exposure to print; indeed, chronological age has been found to account for variance in reading attainment in other developmental disabilities such as Down syndrome (Boudreau, 2002). This finding is nonetheless considerable, given that deficits in related language domains, such as oral (i.e., expressive) language, persist even when older children with FXS are compared to younger mental age-matched controls (Roberts, Chapman, & Warren, 2008; Roberts, Hennon, et al., 2007).

Phonological difficulties in FXS persisted even when mental age was controlled, suggesting that cognitive factors alone cannot account for poor performance on phonological tasks in FXS. In particular,

short-term phonological working memory assessed via the Memory for Words subtest was a weakness relative to other phonological skills tested, and the robust effect size (2.39) detected in group comparisons of this domain supports phonological working memory as an area of difficulty for boys with FXS. Weakness relative to the mental age-matched boys with TD was also detected in synthesizing sounds (Sound Blending subtest). Given that a whole-word over phonological-based approach to literacy instruction is often recommended in the education of individuals with FXS (e.g., Fragile X Clinical & Research Consortium on Clinical Practices, 2012), it is possible that the weak phonological skills exhibited by the boys with FXS are related to a lack of formal instruction in this area.

Predictors of Literacy Skills in FXS

Phonological awareness skills predicted performance on three of the four reading tasks, accounting for 11-14% of the variance in reading skills. This is consistent with a number of studies supporting phonological awareness as a key ingredient for reading success in typical development (Ehri, Nunes, Stahl, & Willows, 2001; Ehri, Nunes, Willows, et al., 2001)*,* as well as emerging evidence supporting the efficacy of a phonological-based approach in the literacy instruction of individuals with other intellectual disabilities such as Down syndrome (Browder, Wakeman, Spooner, Ahlgrim-Delzell, & Algozzine, 2006; Cupples & Iacono, 2002; Lemons & Fuchs, 2010). Findings suggest that phonological skills are critical to the reading success of individuals with FXS and highlight the need for intervention studies to evaluate the effectiveness of phonological-based approaches to literacy instruction for individuals with FXS. Findings also indicated that greater ASD symptom severity predicted poorer performance on several of the phonological tasks, although it was not predictive of the reading variables.

Future Directions

This study is the first to examine reading and phonological skills in a relatively large sample of boys with FXS, providing a starting point for understanding profiles and predictors of literacy ability in this population. Several methodological considerations should be taken into account when interpreting the results of the present study. First, ASD characterization in future work might be strengthened through the use of standardized autism severity metrics, such as that provided by the Autism Diagnostic Observation

Scale (Lord, Rutter, DeLavore, & Risi, 2001). Although the CARS total score has been widely used as a continuous measure of ASD symptoms (e.g., Bailey et al. 2001, Denmark et al., 2003, Hatton et al., 2009, Hatton et al., 2006; Sullivan et al., 2007), raw totals may be more influenced by participant demographics, such as IQ, than are calibrated severity metrics (Gotham, Pickles, & Lord, 2009). Secondly, potential floor effects may have influenced findings, particularly considering the difficulty in identifying literacy measures that are sensitive to variation among individuals with significant intellectual disabilities. However, the use of W scores strengthened our ability to detect variability, as W scores are on an equal interval scale (unlike age equivalent scores) and are norm-referenced (unlike raw scores), allowing for more robust statistical modeling.

It is also notable that participants with FXS were older than their mental age-matched peers, and therefore presumably had greater exposure to print and more years of formal literacy instruction; the observed literacy strengths in the group with FXS may be attributable, in part, to their older age. The choice of comparison group is a challenge inherent to intellectual disabilities research. The inclusion of younger, mental age-matched typically developing controls permit matching on relevant cognitive domains, although this approach inevitably leads to a mismatch in the chronological age of the groups (e.g., Hodapp & Dykens, 2001). The inclusion of comparison groups comprised of children with other developmental disabilities is a method that may be utilized in future work to circumvent potential age confounds, as this strategy allows for both mental-age and chronological-age matching within the same sample.

Important next steps of this work include investigation into the types of literacy instruction received by individuals with FXS in schools; anecdotally, our clinical experience suggests that there is great variability in the types of literacy instruction received by individuals with FXS, as well as variability in related factors such as classroom type, teacher training, and the home literacy environment. There is also a critical need for intervention research to determine the relative effectiveness of phonics versus whole-word literacy instruction for children with FXS. Phonetics-based approaches have proven successful for other developmental disability groups, including populations such as Down syndrome who

have known relative weaknesses in phonological skills(e.g., Conners et al., 2005; Cupples & Iacono, 2002). For instance, in an intriguing intervention study, Cupples and Iacono (2002) found that although the reading skills of children with Down syndrome improved with both phonetics- and whole-word-based instructional approaches, only those children who were taught with the phonetics-based approach were able to generalize skills to reading novel words. Notably, the children with Down syndrome benefited from phonetics-based instruction, *despite* documented weaknesses in phonological skills (indexed by poor auditory working memory ability). Thus, phonological weaknesses do not necessarily preclude children from benefiting from phonetics-based instruction, and the findings of the present study suggest that a phonetics-based approach may be indicated for children with FXS as well. In summary, the present study demonstrates that individuals with FXS are capable of achieving reading and phonological skills, underscoring the importance of targeted, evidence-based literacy instruction for individuals with FXS.

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Integrity of Research and Reporting

Study procedures were approved but the Institutional Review Board of the XX. Informed consent was obtained prior to inclusion of the study.

Conflict of Interest

The authors declare that they have no conflict of interest.

Table 1

Participant characteristics

	Group		Test of Group Differences
	FXS *n* = 51	TD *n* = 35	*p*-value
Chronological Age (years)			.001
M (*SD*)	10.2 (1.7)	5.1 (0.8)	
Range	7.9–13.2	3.3–7.4	
Nonverbal Mental Age[1] (years)			.628
M (*SD*)	5.4 (0.6)	5.2 (0.8)	
Range	4.1–6.7	3.7–7.5	
IQ[2]			.001
M (*SD*)	56.0 (10.47)	107.7 (8.77)	
Range	36.0-74.0	85.0-123.0	
CARS			n/a
M (*SD*)	27.5 (4.5)	--	
Range	17.5–37.5		
Ethnicity [% (*n*)]			.723
European American	82.3 (42)	88.5 (31)	
African American	13.7 (7)	8.6 (3)	
Hispanic	2.0 (1)	2.9 (1)	
Asian American	2.0 (1)	--	
Maternal Education [% (*n*)]			.003
High school or less	20.0 (39.2)	4.0 (11.4)	
Some college	18.0 (35.3)	11.0 (31.4)	
College degree or higher	11.0 (21.6)	20.0 (57.2)	
Not reported	2.0 (3.9)	--	

Note. [1]Brief IQ Age equivalent on the Leiter International Performance Scale-Revised; [2]Brief IQ Standard Score on the Leiter International Performance Scale-Revised; CARS = Childhood Autism Rating Scale

Table 2

Descriptive statistics of group performance on the reading measures

Group	WJ-R Subtest							
	Letter-Word Identification		Passage Comprehension		Word Attack		Broad Reading Composite	
	W scores	Standard scores	W scores	Standard scores	W scores	Standard scores	W scores	Standard scores
FXS	*n* = 51		*n* = 51		*n* = 49		*n* = 51	
M (*SD*)	398.8 (31.2)	45.9 (18.6)	409.5 (39.2)	44.0 (19.1)	440.8 (11.0)	57.6 (13.0)	404.1 (27.8)	41.1 (19.0)
Range	335-450	3-84	380-477	9-95	436-479	40-91	362-464	2-86
TD	*n* = 35		*n* = 32		*n* = 32		*n* = 35	
M (*SD*)	391.0 (20.6)	104.9 (12.4)	394.7 (21.2)	113.7 (25.9)	440.4 (10.5)	104.2 (30.3)	392.9 (17.9)	113.2 (22.0)
Range	356-450	82-133	350-470	88-200	436-477	81-200	368-460	89-183

Table 3

Descriptive statistics of group performance on the phonological measures

	WJ-III Subtest									
Group	Sound Blending		Incomplete words		Memory for Words		Rapid Picture Naming		Phonemic Awareness Composite	
	W scores	Standard scores	W scores	Standard scores	W scores	Standard scores	W scores	Standard scores	W scores	Standard scores
FXS	*n* = 51		*n* = 50		*n* = 51		*n* = 49		*n* = 48	
M (*SD*)	473.6	65.2	486.8	78.1	421.7	43.2	468.9	67.0	482.1	69.0
	(21.2)	(25.2)	(17.5)	(25.7)	(20.0)	(14.4)	(18.4)	(20.2)	(16.1)	(24.4)
Range	430-512	11-116	438-515	1-119	378-470	12-80	431-507	30-107	448-507	5-117
TD	*n* = 35		*n* = 35		*n* = 34		*n* = 35		*n* = 35	
M (*SD*)	488.7	115.6	493.0	115.5	467.6	104.8	471.9	102.2	490.7	119.7
	(10.7)	(16.8)	(9.3)	(13.5)	(20.7)	(11.5)	(12.9)	(11.9)	(8.1)	(12.1)
Range	472-528	97-186	478-515	87-150	426-507	82-134	442-498	81-127	476-511	95-154

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