

# Linguistic and Cognitive Functioning and Autism Symptoms in Young Children With Fragile X syndrome

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## Abstract

Linguistic and cognitive profiles were examined in 18 children with autism and 18 children with fragile X syndrome (mean ages = 34 months). State-of-the-art diagnostic procedures for autism symptom identification were administered. Eight children with fragile X met criteria for autism. Comparison of linguistic and cognitive profiles (autism, fragile X without autism, fragile X with autism) revealed that children with fragile X (with autism) were more impaired in nonverbal cognition and expressive language. Receptive language was a relative strength for children with fragile X (without autism). There were no differences in receptive language in children with autism, regardless of fragile X status. Low receptive language may be a marker for autism symptoms in young children with fragile X.

Fragile X syndrome is a genetic disorder associated with mental retardation, specific physical features, language delays, and a characteristic behavioral phenotype. This syndrome is currently regarded as the most common inherited cause of mental retardation (Hatton, Bailey, Hargett-Beck, Skinner, & Clark, 1999), and current prevalence estimates are 1:3,200 to 4,000 in males and 1:6,000 in females (Sherman, 2002). Variability in behavioral and cognitive characteristics is often noted, and a general cognitive decline has been documented across late childhood and early adolescence in studies of males with fragile X (as reviewed in Bennetto & Pennington, 1996). Behavioral characteristics frequently reported include social avoidance or anxiety, hyperarousal to sensory stimuli, distractibility, irritability, high activity level, repetitive motor behaviors, and difficulty coping with changes and unpredictable events (Bailey, Hatton, & Skinner 1998; Hatton et al., 1999).

In addition, specific speech and language features are associated with fragile X syndrome (Abbeduto & Hagerman, 1997; Paul, Cohen, Breg, Watson, & Herman, 1984). To date, the vast majority of speech–language researchers have focused on atypicalities of spoken or expressive language, generally by considering the language characteristics of adults and adolescents with fragile X (Dykens, Hodapp, & Leckman, 1994; Hagerman & Cronister, 1996; Sudhalter & Belser, 2001; Wolf-Schein et al., 1987). Delayed acquisition of functional speech, repetitive speech, dysfluency, misarticulations, and unusual prosodic features have been reported (Abbeduto & Hagerman, 1997; Dykens et al., 1994; Turner, Daniel, & Frost, 1980; Wolf-Schein et al., 1987). Newell, Sanborn, and Hagerman (1983) reported that relative strengths in receptive language abilities are often associated with X-linked retardation syndromes, and, in fact, there is some evidence that children with fragile X demonstrate relatively better receptive than ex-

pressive abilities (Paul et al., 1984). Receptive language ability increases more quickly compared to expressive language during early childhood in most males with fragile X (Roberts, Mirret, & Burchinal, 2001; Paul, Dykens, Leckman, Watson, & Herman, 1987). Furthermore, by adolescence, receptive language has been reported to be mental age (MA) appropriate in boys with fragile X syndrome who do not meet criteria for an autism spectrum disorder (Abbeduto et al., 2003). Roberts, Boccia, Bailey, Hatton, and Skinner (2001) suggested that this splintered profile of language development characterizes the communicative aspect of the behavioral phenotype of fragile X syndrome; however, their evidence suggests that the split in receptive and expressive skills does not emerge until after 66 months, with discrepancies increasing with age.

There is quite a debate concerning whether autism symptoms in fragile X syndrome are reflective of a common etiological pathway or an artifact of a significant cognitive delay. Although no large epidemiological studies have been conducted on the comorbidity of fragile X and autism, some researchers using relatively small clinical samples have suggested a comorbidity rate of approximately one fourth to one third of children with fragile X meeting behavioral criteria for autism (Bailey et al., 1998; Rogers, Wehner, & Hagerman, 2001; Turk & Graham, 1997). Researchers concur on the heavy preponderance of “autistic-like” symptoms inherent in a diagnosis of fragile X syndrome (Brown et al., 1986; Cohen, Sudhalter et al., 1991; Hagerman, Jackson, Levitas, Rimland, & Braden, 1986). Behaviors observed in children with fragile X that resemble behaviors seen in children with autism include avoidance of eye gaze, unusual reactions to environmental stimuli, hand stereotypies, hand-biting, spinning, and inappropriate and repetitive use of objects (Levitas, Braden, Van Norman, Hagerman, & McBogg, 1983). Furthermore, several of the language characteristics observed in persons with fragile X are also observed in individuals with autism, such as delayed expressive language development (Paul et al., 1987), poor social use of language (Newell et al., 1983), unusual prosody, tangential speech, and repetitive language (Wolf-Schein et al., 1987).

By contrast, there are some significant differences in cognitive and language skills in persons with autism and those with fragile X. Unlike most children with fragile X syndrome, children with autism tend to have significant recep-

tive language delays relative to other skills, which have been likened to those observed in developmental receptive dysphasia (Bartak, Rutter, & Cox, 1977; Jarrold, Boucher, & Russell, 1997; Konstantareas & Beitchman, 1996; Rapin, 1996; Tager-Flusberg & Sullivan, 1998). By contrast, nonverbal problem-solving tends to be a relative strength in autism (Bauman, 1999; Rapin, 1991, 1996). Consideration of these findings suggests that examining the early cognitive and language profiles of young children with fragile X syndrome, and their relation to autism symptoms, may provide some clues as to how these conditions are similar or different.

Symptom overlap does not necessarily imply common etiologies (Bailey, Hatton, Skinner, & Mesibov, 2001; Cohen, 1995). Lower functioning children with fragile X may, in fact, look autistic at an early age, as a result of considerably delayed development (Bailey et al., 2001; Rogers et al., 2001). Supporting this argument is the ample evidence that autism symptoms are more commonly reported in older children, adolescents, and adults who are lower functioning (Bailey, Mesibov et al., 1998; Bailey et al., 2001; Cohen, Sudhalter et al., 1991; Feinstein & Reiss, 1998; Hagerman et al., 1986; Reiss & Freund, 1990; Rogers et al., 2001). In addition, important differences in language functioning of conversational males with fragile X and those with autism suggest that there is less symptom overlap between autism and fragile X syndrome in higher functioning persons (Sudhalter & Belser, 2001; Sudhalter, Cohen, Silverman, & Wolf-Schein, 1990).

One way to evaluate the relation between fragile X and autism is to examine autism symptoms as well as linguistic and cognitive functioning in very young children with fragile X syndrome, using state-of-the-art diagnostic measures. This is what we proposed for this study. We expected that the initial state of a behavioral phenotype will be less affected by developmental processes than a later manifestation of the same phenotype (Oliver, Johnson, Karmiloff-Smith, & Pennington, 2001) and, thus, be more informative about the core features. Given the equivocal nature of the co-morbidity issue in fragile X syndrome and autism, there are two possible hypotheses for this study: (a) Do young children with fragile X syndrome who meet criteria for autism demonstrate a different pattern of early linguistic and cognitive skills compared to young children with fragile X syndrome who do not meet criteria

for autism? (b) Do young children with fragile X syndrome who meet criteria for autism demonstrate a similar pattern of early linguistic and cognitive skills compared to young children with autism only (i.e., a weakness in receptive language relative to nonverbal problem-solving ability)?

If the first hypothesis is correct, then we may assert that “classic” autism truly occurs within fragile X syndrome. If the second hypothesis is correct, then it is more likely that developmental status is influencing social and communicative functioning. Identification of early markers of autism symptoms in children with fragile X syndrome and examination of symptom expression across development will help to inform this debate.

## Method

### Participants

Thirty-six participants were included and comprised two groups as a function of diagnosis: autistic disorder ( $n = 18$ ) and fragile X syndrome ( $n = 18$ ). All children were males between the ages of 26 and 45 months. There were no significant differences between these two groups on nonverbal mental age (MA) or chronological age (CA). Participant characteristics are shown in Table 1.

All participants were recruited from various health and early education agencies as well as parent support groups (e.g., Fragile X Foundation and Autism Society of America). The children with fragile X were recruited from a national parent network and specialty fragile X clinics across the country (primarily in Denver, CO; Oakland, CA; and Chapel Hill, NC). The groups were quite similar in ethnic distribution and socioeconomic status—SES (Hollingshead, 1975).

The children with autism were free from any other medical condition (including a diagnosis of

fragile X syndrome), had no visual or hearing impairment, had been diagnosed with autism by an outside agency, received current clinical diagnoses of autism by psychologists with an expertise in autism, and met criteria for autism on three diagnostic measures: Diagnostic and Statistical Manual-IV—DSM-IV checklist (American Psychiatric Association, 1994), Autism Diagnostic Interview-Revised (Lord, Rutter, & LeCouteur, 1994), and Autism Diagnostic Observation Schedule-Generic (Lord, Rutter, DiLavore, & Risi, 1999). All but 4 of the participants with autism received negative testing for fragile X syndrome. Parents of the remaining 4 participants, who were not tested, refused testing despite clinical encouragement. Nonetheless, these 4 participants were not showing or suggesting evidence of a diagnosis of fragile X syndrome. (See below for descriptions of these measures.)

Diagnosis of fragile X was based upon molecular genetic testing, which was completed prior to the participants' enrollment in this study. The children with fragile X were free from other medical condition(s) and had no visual or hearing impairment. In order to identify children with comorbid autism, we also administered an intensive diagnostic protocol to participants with fragile X. Eight participants with fragile X met criteria for autism on three diagnostic systems (i.e., DSM-IV, Autism Diagnostic Interview-Revised, and Autism Diagnostic Observation Schedule) as well as clinical judgment by a psychologist with an expertise in autism. These participants comprised the fragile X with autism subgroup. The rest of the fragile X participants did not meet criteria on more than one of the three diagnostic systems and comprised the fragile X (without autism) group. One participant in this group met criteria for autism on the Autism Diagnostic Interview-Revised only, and 1 partic-

**Table 1.** Participant Characteristics by Diagnostic Group ( $N = 36$ )

Variable	Autism ( $n = 18$ )			FXS <sup>a</sup> without autism ( $n = 10$ )			FXS with autism ( $n = 8$ )		
	Mean	SD	Range	Mean	SD	Range	Mean	SD	Range
CA (months)	34.28	3.4	26–41	34.80	7.4	22–45	36.00	5.3	27–45
ADOS <sup>b</sup> total score	17.53	4.18	8–23	5.10	2.92	1–10	19.13	3.9	15–24
ADI <sup>c</sup> total score	33.28	6.2	19–43	18.60	6.2	8–29	31.28	5.5	22–39
SES (Hollingshead)	50.66	9.8	29–66	50.93	13.4	15–66	57.10	11.0	38–66

<sup>a</sup>Fragile X syndrome. <sup>b</sup>Autism Diagnostic Observation Schedule. <sup>c</sup>Autism Diagnostic Interview.

ipant met criteria on the Autism Diagnostic Observation Schedule only. These participants did not meet DSM-IV criteria for autism and were determined by clinical judgment not to meet criteria for autism. Thus, they were included in these analyses in the fragile X (without autism) subgroup because (a) they are representative of the children seen by clinicians and (b) any bias that would be introduced would be a conservative one.

*Closer look at autism symptoms.* We also examined symptoms of autism in more detail in order to see whether the behavioral phenotype of autism in fragile X is distinct from that of autism without fragile X. In order to examine which symptoms tend to differentiate autism within the fragile X group, we reduced Autism Diagnostic Interview-Revised items to dichotomous scores (0 = no impairment, 1 = some or severe impairment, as evidenced by a score of 1, 2, or 3). Due to the number of comparisons, we set alpha at .01. Children with comorbid autism demonstrated many communication and social difficulties but were not significantly different in play or other behaviors from children with fragile X without autism (see Table 2).

### *Developmental Measures*

*Mullen Scales of Early Learning (Mullen, 1995).* The Mullen Scales, a standardized developmental test for children ages 3 months to 60 months, consists of five subscales: Gross Motor, Fine Motor, Visual Reception, Expressive Language, and Receptive Language.

This instrument allows for separate standard verbal and nonverbal summary scores to be constructed and demonstrates strong concurrent validity with other well-known developmental tests of motor, language, and cognitive development (Mullen, 1995). The Mullen Scales was administered to all participants according to standard instructions by raters with advanced degrees, trained in assessing young children with autism and other developmental disorders. Reinforcers (e.g., food, social praise) for all participants in all groups were used at times to reward cooperation and attention. Nonverbal developmental quotients were calculated as follows: (visual-receptive age equivalent/CA)  $\times$  100. Receptive and expressive developmental quotients were calculated in the same manner. We decided to use developmental quotients rather than standard scores in an effort to avoid floor effects.

**Table 2.** Comparison of Autism Diagnostic Observation Schedule Items Between Fragile X Subgroups (With and Without Autism)

Item	$\chi^2$
Overall level of nonechoed language	15.99
Frequency of vocalizations directed to others	29.73**
Intonation	14.03**
Immediate echolalia	5.63
Pointing	13.44
Gestures	14.71
Idiosyncratic words	3.94
Use of other's body to communicate	3.77
Unusual eye contact	26.56**
Showing	20.34**
Facial expression directed towards others	19.31**
Initiation of joint attention	12.78**
Response to joint attention	17.73**
Social overtures	30.44**
Functional play	18.16**
Imagination/creativity	14.81
Sensory interests	9.35
Hand and finger mannerisms	5.31
Self-injurious	5.62
Overactivity	6.44

\*\* $p < .01$ .

We used the Receptive and Expressive Language Scales (Mullen, 1995) to measure overall language functioning in our participants in order to compare developmental profiles; thus, more specific areas within language domains (e.g., semantics/vocabulary, pragmatics/social use of language, syntax/grammar) were not assessed. The Receptive Language Scale provides an assessment of a child's ability to decode verbal input. The majority of questions require a nonverbal response (such as pointing or performing a specific command), with the exception of one higher-level item, which requires a child to answer questions in order to ascertain their general knowledge skills (e.g., What do we wash our hands with? How many legs does a horse have? Why do we have refrigerators?). The Expressive Language Scale assesses a child's spontaneous language, specific vo-

cal or verbal responses to questions, and higher-level concept formation (Mullen, 1995).

### *Autism Symptoms*

*Autism Diagnostic Interview-Revised.* This instrument is a structured, standardized parent interview developed to assess the presence and severity of symptoms of autism in early childhood across all three main symptom areas involved in autism: social relatedness, communication, and repetitive behaviors. The Autism Diagnostic Interview-Revised has been carefully validated across a wide range of ages and severity levels in autism. An algorithm has been established that differentiates autism from other developmental disorders at high levels of sensitivity and specificity (over .90 for both) for participants with MAs of 18 months and older. One of the developers of the instrument (Lord) trained the last author to reliability on the Autism Diagnostic Interview-Revised, who then trained other raters in the lab to reliability of 80% or better item agreement on three consecutive administrations using the full range of scores (0 to 3) rather than the truncated scoring usually used (0 to 2). Reliability was maintained at 80% for 20% of participants across the period of data gathering.

*Autism Diagnostic Observation Schedule–Generic.* The Autism Diagnostic Observation Schedule is a semi-structured standardized observation using developmentally appropriate social and toy-based interactions in a 30- to 45-minute interaction to elicit symptoms of autism in four areas: social interaction, communication, play, and repetitive behaviors. This instrument consists of four modules, each directed at a particular level of language ability. In the present study, all participants received Module 1, which was designed for preverbal children or those just beginning to speak.

The Autism Diagnostic Observation Schedule and its predecessors, the Autism Diagnostic Interview-Revised and the Pre-Linguistic Autism Diagnostic Observation Schedule, have been carefully validated across a wide range of ages and severity levels in autism (DiLavore, Lord, & Rutter, 1995; Lord et al., 2000.) In the present study, Lord also trained the final author to reliability on the Autism Diagnostic Observation Schedule, who then trained other raters in the lab to reliability of 80% or better item agreement on three consecutive administrations using the full range of scores (0 to 3). Reliability was maintained at 80% for 20% of participants across the period of data gathering.

### *Procedure*

This study was part of a larger, longitudinal study of the developing phenotype of autism and fragile X syndrome. The entire study was carried out under Institutional Review Board (IRB) approval. Consent forms were reviewed with each family, and all questions were answered before consent was obtained and before any measures were gathered. Mothers participated in the Autism Diagnostic Interview-Revised generally during a home visit. The Autism Diagnostic Observation Schedule and Mullen Scales of Early Learning were administered in the lab over two visits, which generally occurred within a month's time, along with other measures not reported here.

## Results

### *Preliminary Analyses*

Our first step was to examine item-by-item performance in the autism and fragile X groups in order to ascertain whether the Mullen Scales of Early Learning Language Scales represents a developmental sequence that is meaningful for young children demonstrating atypical development. Our concern was that if young children with various developmental disabilities demonstrate an atypical progression of language acquisition on the Mullen Scales, then statistical analysis of group differences on overall scores would be of questionable validity. A variation of the Guttman scalogram analysis (refer to Green, 1956) was performed on a selected set of items (for children ages 7 to 33 months) based upon where the majority of participants were obtaining their basal and ceiling scores. Summary statistics were used to evaluate the pattern of skill acquisition within each language domain for each group. The items are considered “scalable,” or progressing in difficulty, if  $I$  is more than .50. Scores lower than .50 indicate that these items do not form a coherent scale.

Nonverbal Problem-Solving, Receptive, and Expressive Language scales were found to be scalable (or appropriately sequenced developmentally) for all groups: autism:  $I$  (Nonverbal) = .88,  $I$  (Receptive) = .72,  $I$  (Expressive) = .70; fragile X (with autism):  $I$  (Nonverbal) = .89,  $I$  (Receptive) = .93,  $I$  (Expressive) = .77; fragile X (without autism):  $I$  (Nonverbal) = .87,  $I$  (Receptive) = .70,  $I$  (Expressive) = .67. Therefore, we can conclude that the scales of the Mullen Scales of Early Learn-

ing are appropriate measures for these young children with atypical patterns of development.

Prior to conducting group comparisons, we examined the distribution of scores within each group. Variances in age equivalent scores were comparable across all groups in nonverbal, Levene’s  $F(3, 32) = .10, p = .98$ , receptive, Levene’s  $F(3, 32) = .07, p = .48$ , and expressive, Levene’s  $F(3, 32) = .70, p = .56$ , functioning. Distributions were fairly normal for the children with autism and fragile X without autism; however, the distributions of both receptive and expressive language functioning were negatively skewed for the children with comorbid fragile X and autism. Due to floor effects evident in the standardized scores, we computed a developmental quotient for the language variables using the following formula:  $(MA\text{-equivalent}/CA) \times 100$ . These developmental quotients were used in subsequent analyses.

*Group Comparisons of Cognitive Functioning by Diagnosis*

A MANOVA was used in which the dependent measures were the variables reflecting the three levels of cognitive domain: nonverbal, receptive, and expressive developmental quotients. The between-group factor was diagnosis, which also has three levels: autism, fragile X syndrome without autism, and fragile X syndrome with autism. There were significant effects for cognitive domain: Wilks Lambda,  $F(2, 32) = 31.67, p = .000, \epsilon^2 = .66$ ; and for diagnostic group: Wilks Lambda,  $F(2, 33) = 12.53, p = .000, \epsilon^2 = .43$ . There was also a significant interaction between cognitive domain and diagnostic group,  $F(4, 64) = 6.37, p = .000, \epsilon^2 = .29$  (see Table 3).

Results suggest that children with fragile X and autism were lower functioning in nonverbal and expressive language functioning than children with either autism or fragile X syndrome. More interesting is the finding that children who have fragile X syndrome, but do not meet criteria for autism, obtained significantly higher receptive language scores than did other children in the study, suggesting that receptive language ability may be a marker for autism symptoms in fragile X syndrome.

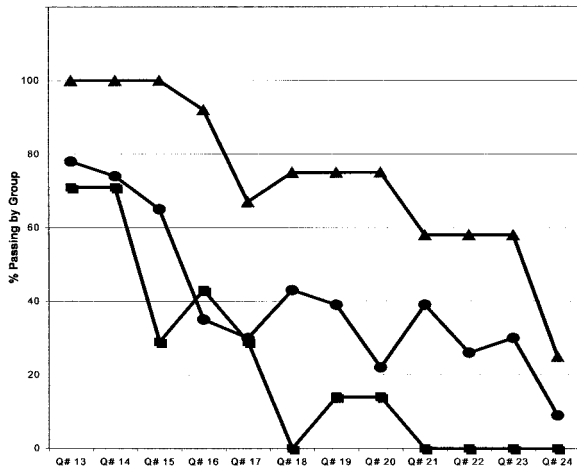
Due to the small number of participants in the fragile X subgroups, we also conducted non-parametric analyses to address the same question: Is there a difference in pattern of cognitive functioning by diagnostic subgroup? We used a Kruskal-Wallis test and obtained significant differences in rankings by group across all three domains of cognitive function, such that children with fragile X and autism were lower functioning than the other two groups in nonverbal and expressive functioning but were not different from those with autism in receptive language functioning: nonverbal developmental quotient,  $H(1, 32) = 9.67, p < .01$ ; receptive developmental quotient,  $H(1, 32) = 12.00, p < .01$ ; and expressive,  $H(1, 32) = 8.08, p < .01$ .

A closer look at the performance on items in the 12- to 24-month age range by diagnostic group is presented in Figure 1. This age range was chosen because it allowed us to best explore where the children with the lowest receptive language skills were reaching their ceiling. Visual inspection of these data suggests that although the participants with fragile X (with autism) group performed more poorly than did members of either

**Table 3.** Means and SDs of Groups on Cognitive Domains With MANOVA

Variable <sup>a</sup>	Group						Comparisons ( <i>N</i> = 36)	
	1 Autism ( <i>n</i> = 18)		2 FXS <sup>b</sup> without autism ( <i>n</i> = 10)		3 FXS with autism ( <i>n</i> = 8)			
	Mean	SD	Mean	SD	Mean	SD	<i>F</i> (2, 32)	Contrast <sup>c</sup>
NVDQ	63.9	12.7	64.3	9.1	43.1	9.3	10.98*	3 < 1*, 2*
RLDQ	43.7	20.9	72.3	16.1	31.6	8.9	13.35*	1 < 2*; 3 < 2*
ELDQ	43.4	16.4	46.6	20.5	18.6	9.1	7.9	3 < 1*, 2*

<sup>a</sup>NVDQ = Nonverbal Developmental Quotient, RLDQ = Receptive Language Developmental Quotient, ELDQ = Expressive Language Developmental Quotient. <sup>b</sup>Fragile X syndrome. <sup>c</sup>NVDQ-FXS without autism better than other two groups, RLDQ-FXS without autism better than other two groups, ELDQ-FXS with autism worse than other two groups. \**p* < .05.



**Figure 1.** Receptive language items on the Mullen Scales of Early Learning. Q# 13 = understands commands with gestures. Q# 14 = identifies 1 object (out of 2) on the table. Q# 15 = Follows command “Give me” without a gesture. Q# 16 = comprehends questions about objects in the room without gestures (e.g., Where’s the door?). Q# 17= follows simple one-step directions without a gesture (e.g., Give the block to mommy). Q# 18 = identifies body parts on a pictured boy. Q# 19 = identifies a picture from a field of 3. Q# 20 = follows a related 2-step command (e.g., Stand up and get the ball). Q# 21 = identifies a picture from a field of 4. Q# 22 = shows auditory spatial awareness (e.g., in, on, under, etc.). Q# 23 = comprehends action words in a picture (e.g., eating, washing, sleeping). Q# 24 = identifies object by its function. The oval represents the autism group; the rectangle, the fragile X (with autism) group; and the triangle, the fragile X (without autism) group.

of the other groups, their performance appears more similar to the autism group than to the fragile X (without autism) group. More participants would be required to examine the finding using inferential statistics.

**Discussion**

This study provides a downward extension of previous work concerning linguistic and cognitive profiles in children with fragile X (Abbeduto et al., 2003; Roberts, Mirret, & Burchinal, 2001) and extends this work to include comparisons with autism. These findings are consistent with previous reports of a relative strength in receptive language

compared to expressive language for children with fragile X who do not have autism. This characteristic profile emerges early and is not apparent in children who are presenting with fragile X and autism. Perhaps autism and fragile X affect language in different ways, with a compounded effect seen in children who are diagnosed with both conditions (see also Bailey et al., 2000). These findings also corroborate Rapin’s (1996) suggestion that deficits in comprehension (receptive language) most strongly differentiate autism from nonautism. Thus, from these comparisons, evidence of unexpectedly poor receptive language in a young child with fragile X may be indicative of a possible autism diagnosis and, therefore, warrants further clinical investigation.

One of the strengths of this study is the methodology used to diagnose autism. All children participated in the Autism Diagnostic Observation Schedule, and an Autism Diagnostic Interview-Revised was administered to the caregiver(s) of every child. Clinicians observed and interacted with the child, interviewed the parents, and reviewed developmental and historical data before making a clinical diagnosis. For the children with fragile X, two experienced clinicians reviewed the data prior to determining a diagnosis on the autism spectrum. We do not yet understand the directionality of the effects observed: Does having autism affect overall development or does overall development create a behavioral picture that looks like autism? Of the 8 children in the fragile X (with autism) group, 6 (86%) presented with overall MAs of 18 months or less, which certainly affects the pattern of social, communicative, and behavioral functioning observed in these children. Only 3 children (24%) with fragile X (without autism) presented with MAs below 18 months. Furthermore, of critical importance in determining the nature of the early expression of autism in fragile X syndrome, comparison of nonverbal skills to the linguistic profile was necessary. In this study, the subset of young children with a comorbid diagnosis of fragile X and autism showed significant impairment across all domains of developmental functioning, suggesting that nonverbal skills are not a relative strength and that overall developmental functioning is severely impacted by the dual diagnosis. This finding most closely supports our hypothesis that the lower developmental status seems to be influencing the development of social and communication skills. “Classic” autism

does not appear to be occurring in these young children with fragile X syndrome.

In terms of the language profiles of young children with autism (without fragile X), we found a flat linguistic profile, with no apparent discrepancies between expressive and receptive language abilities. This is different from the studies on older children with autism, in which receptive abilities are reported to be an area of relative weakness for some children (Rapin, 1996). Perhaps relative impairments in language understanding appear to emerge over time as developmental expectations in this domain increase across childhood. Alternatively, perhaps children with autism acquire expressive skills in order to meet instrumental needs, but appear to lag in the development of receptive abilities due to impairments in core social relating. Learning to communicate a simple message to get one's needs met (emerging expressive language) may be more powerful and functional for a child who is less attuned to the social aspects of communication than learning to respond to others' communications (emerging receptive language). Considering that some of the earliest symptoms observed in young children with autism include a lack of social orienting (Baranek, 1999; Osterling & Dawson, 1994), acting as if deaf, and failure to respond when name is called (Dahlgren & Gillberg, 1989; Lord et al., 1999; Ohta, Nagai, Hara, & Sasaki, 1987), perhaps impairments in receptive language are early indicators of impairments in core social relatedness.

Results of previous research suggest that deficits in core social relatedness are key for differentiating autism in children with fragile X (Rogers et al., 2001). Although children with fragile X may be socially anxious or even avoidant, early dyadic interactions with familiar caregivers (including sharing affect, playing simple social games) can be quite rich, suggesting that the social impairment in fragile X is not in core social relatedness, but perhaps in social anxiety or hyperarousal (Hagerman & Hagerman, 2002; Roberts, Boccia et al., 2001). Conversely, for those children with comorbid fragile X and autism, core social relatedness appears to be significantly affected, based upon diagnostic testing and clinical observation, and impairments in receptive language, rather than avoidance, may be reflective of this limited capacity.

Overall, these findings have specific implications for guiding interventions in the area of language development for children with fragile X

syndrome. Other authors have suggested that children with different behavioral phenotypes benefit from different intervention approaches, based upon specific profiles of strengths and weaknesses (Dykens & Hodapp, 2001; Hodapp & Fidler, 1999). For example, young children with autism may need more interventions focused upon building receptive skills, whereas children with fragile X may derive more benefit from interventions targeting expressive skills. Children with comorbid fragile X and autism may need intense intervention in both areas. In addition, recognizing the relative strengths in receptive language in young children with fragile X has many implications for educational and treatment strategies, such as providing a therapist and child with a common point of understanding in order to furnish rich and meaningful opportunities to learn and practice expressive activities.

These findings also have implications for assessment and evaluation practices. Inspection of the language profile of young children during diagnostic evaluations might help to screen for autism, particularly in a case where a child presents with fragile X. Although tests for fragile X are objective and definitive, testing for autism is based on behavior and can be more difficult to ascertain, especially for professionals lacking clinical experience with this population. The complexity of diagnosis is compounded when a child has fragile X, a syndrome that presents with several features that may seem similar to those detected in autism. Given the finding that receptive language skills may distinguish children with autism from children with fragile X, children with fragile X who present with an even and severely depressed linguistic profile should be assessed for possible autism. Receptive language skills may provide important information for differential diagnosis of autism and fragile X.

There are several limitations to this study that must be kept in mind when considering the findings. First, the sample sizes were small, particularly in the fragile X group. Larger samples would yield stronger findings that could be more easily generalized to the populations under consideration. Second, our autism and fragile X study groups were limited to males; thus, the findings cannot be generalized to young girls with these disorders. Finally, until we are able to observe these children in follow-up visits, we cannot comment on the stability of the autism symptoms within the fragile X group.

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