Research Article

Gesture Frequency and Function in Infants With Fragile X Syndrome and Infant Siblings of Children With Autism Spectrum Disorder

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Purpose: Infant siblings of children with autism spectrum disorder (ASIBs) and infants with fragile X syndrome (FXS) are both at risk for developing autism spectrum disorder (ASD) and communication disorders; however, very few studies have examined 1 of the earliest forms of intentional communication in infants from these groups: gestures. This study examined the frequency and function of gesture use across 12-month-old infant ASIBs, infants with FXS, and low-risk controls.

Method: Participants included 23 ASIBs who did not later meet diagnostic criteria for ASD, 18 infants with FXS, and 21 low-risk controls. Gestures were coded from a semistructured play-based interaction.

Results: Overall, infants with FXS displayed fewer gestures than low-risk infants, whereas ASIBs did not

differ from the FXS or low-risk groups in overall gesture frequency. In terms of the communicative function of the gestures used, the FXS and ASIB groups displayed significantly fewer social interaction gestures than the low-risk controls, with large effect sizes.

Conclusion: This study contributes to scant knowledge of early communication phenotypes of infant ASIBs who do not meet criteria for ASD and infants with FXS. Results indicated that gesture function, not frequency, best discriminated at-risk infants from low-risk infants at 12 months of age. Findings have implications for the clinical evaluation and treatment of infants at high risk for ASD and communication disorders.

Gestures emerge as early as 6–9 months of age and represent one of the first forms of intentional communication (Crais, Douglas, & Campbell, 2004). Gestures almost always precede the use of verbal language (Parladé & Iverson, 2011) and are closely tied to later vocabulary development and expressive language ability (Rowe, Özçaliskan, & Goldin-Meadow, 2008). Therefore, gestures in infancy can serve as one of the earliest markers for atypical communication development, even before delays in spoken language become apparent (Crais, Watson, & Baranek, 2009). Examining gestures may be particularly important in the early identification of neurodevelopmental disorders such as autism spectrum disorder

(ASD) and fragile X syndrome (FXS), which are characterized primarily by social communication deficits. Infant siblings of children with ASD (ASIBs) and infants with FXS are both at elevated risk for ASD and related communication difficulties, but few studies have examined early gesture development within these populations. The unique characteristics of each group predispose infants to distinct communicative profiles; therefore, it is important for clinicians to be well informed of syndrome-specific profiles in order to identify early delays and plan effective language and communication intervention.

Risk for Nontypical Outcomes in ASIBs

The diagnosis of ASD is based on a series of behavioral deficits, with social-communicative impairment as one of the most ubiquitous (American Psychiatric Association, 2013). Although the diagnosis of ASD is determined via behavioral symptoms, evidence supports a strong genetic basis to the condition (e.g., De la Torre-Ubieta, Won, Stein, & Geschwind, 2016). Younger siblings of individuals diagnosed with ASD (ASIBs) are at elevated genetic risk of developing ASD themselves, with a recurrence rate of about

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20% (Ozonoff et al., 2011). Of those ASIBs who do not meet diagnostic threshold for ASD (i.e., non-ASD ASIBs), about 40% will be affected by one or more nontypical outcome, such as language delay, global developmental delay, and subthreshold ASD symptoms known as the *broad autism phenotype* (BAP; Charman et al., 2017; Messinger et al., 2013; Ozonoff et al., 2014; Yirmiya et al., 2006). BAP symptoms in non-ASD ASIBs are commonly related to social and communication difficulties, such as lower levels of social smiling, atypical social referencing, reduced eye contact, and lower rates of joint attention (Cassel et al., 2007; Cornew, Dobkins, Akshoomoff, McCleery, & Carver, 2012; Messinger et al., 2013; Nichols, Ibañez, Foss-Feig, & Stone, 2014; Ozonoff et al., 2014; Stone, McMahon, Yoder, & Walden, 2007). However, there is little understanding as to how these differences manifest early in development. In one of the few prior studies on the emergence of nontypical developmental features in non-ASD ASIBs, Ozonoff et al. (2014) found that ASIBs who demonstrated nontypical developmental outcomes at 36 months could be distinguished from low-risk controls on standardized language measures as early as 12 months of age. However, the differences between the non-ASD ASIBs and low-risk controls were most pronounced at older ages, with the groups only showing modest differences on the standardized language measures at 12 months of age and no differences at 6 months (Ozonoff et al., 2014). The analysis of more fine-grained communicative behaviors, such as the use of gestures, may lend deeper insight into the earliest emerging communicative differences in non-ASD ASIBs, as broad-based standardized measures may be insufficient for detecting the earliest emerging communicative differences in non-ASD ASIBs.

FXS as a Condition Associated With ASD Risk

FXS is a genetically based neurodevelopmental disorder that occurs in approximately one in 4,000 males and one in 6,000 females (Sherman, Pletcher, & Driscoll, 2005; Turner, Webb, Wake, & Robinson, 1996). FXS is the most common known single-gene cause of ASD (Auerbach, Osterweil, & Bear, 2011; Darnell et al., 2011). It is caused by a mutation of the *fragile X mental retardation-1* gene, which is located on the X chromosome. The mutation of the *fragile X mental retardation-1* gene inhibits the production of fragile X mental retardation protein, which is essential for brain development (Huber, Gallagher, Warren, & Bear, 2002). Common features of the FXS phenotype include intellectual disability, inattention, hyperarousal, anxiety, and social withdrawal (D. Cohen et al., 2005; Klusek, Roberts, & Losh, 2015; Roberts et al., 2005). Females with FXS tend to display fewer deficits caused by FXS because the second, unaffected X chromosome can compensate for the effects of the mutated X (Lozano, Rosero, & Hagerman, 2014).

Symptoms of ASD are prevalent in FXS; about 60% of males and 20% of females with FXS exhibit enough symptoms to meet diagnostic criteria for ASD (Clifford et al., 2007; Klusek, Martin, & Losh, 2014b). Language

and communication are commonly impaired in FXS, with vocabulary, morphology, syntax, phonology, and literacy skills delayed relative to age-based expectations but generally on par with nonverbal cognitive level (Finestack, Richmond, & Abbeduto, 2009; Klusek, Hunt, et al., 2015). In contrast, social communication and pragmatic language performance in FXS are often more impaired than would be expected relative to cognitive ability and are exacerbated by the presence of ASD symptoms and comorbidity (Abbeduto et al., 2008; Klusek et al., 2014a; Losh, Martin, Klusek, Hogan-Brown, & Sideris, 2012; Roberts et al., 2007). Although communication impairments in FXS are well documented, this work has mostly involved school-age or adolescent individuals, so very little is known about the profile of early communication development.

Understanding the earliest manifestations of communication impairment in infants with FXS can inform the implementation of anticipatory monitoring and early, targeted treatment (e.g., Fidler, Philofsky, & Hepburn, 2007). Additionally, knowledge of communication phenotypes linked with specific genetic etiologies can guide early interventionists in identifying areas in which weaknesses are likely (Fidler et al., 2007). Furthermore, delineating communication profiles linked with various forms of genetic risk can shed light on the role of specific genes in producing certain phenotypes (Dykens, Hodapp, & Finucane, 2000). Direct contrast across etiological groups contributes to these efforts by allowing for the identification of strengths and weaknesses that are shared or distinct across conditions. This study contrasted communicative gesture use in infants with FXS and infant ASIBs who did not receive a later ASD diagnosis. These at-risk groups reflect varying levels and forms of genetic risk for ASD, and therefore, their comparison can inform how distinct etiological risk factors are associated with select communication profiles.

Gesture Development

In typical development, children begin to use gestures between 6 and 9 months of age (Crais et al., 2004; Guidetti & Nicoladis, 2008; Iverson & Goldin-Meadow, 2005). By 12 months of age, varied forms of gestures have emerged and serve a range of communicative functions (Rowe et al., 2008; Watson et al., 2013). The communicative functions of gestures can be broken down into Bruner's (1981) three earliest functions of intentional communication: behavior regulation, social interaction, and joint attention. These functions represent a continuum of sophistication in social behavior, with behavior regulation placed on the low end and joint attention placed on the high end. Behavior regulation gestures first emerge around the age of 6 months, often in the form of protesting, and are used to regulate another person's behavior (Crais et al., 2004; Watson et al., 2013). Examples of behavior regulation gestures may include pushing an object away or reaching to be picked up by another person. Around 8–9 months of age, social interaction gestures emerge and are used to attract a person's attention and engage in social interaction, such as initiating a social

game or routine, teasing, and showing off (Crais et al., 2004). Joint attention gestures, which are used to direct another person's attention to an object or event (e.g., pointing to an object to show shared interest), emerge between 9 and 12 months of age (Crais et al., 2004). Thus, the analysis of gesture function, in addition to frequency, may reveal syndrome-specific communicative patterns in syndromes characterized by social impairment, such as ASD or FXS (Flenthrope & Brady, 2010).

Gestures serve as the building blocks for more advanced communication skills, such as oral language. For example, the frequency and diversity of gestures used in toddlerhood are linked to enhanced vocabulary skills at older ages (e.g., IAcredolo & Godwyn, 1988; Iverson & Goldin-Meadow, 2005; Rowe & Goldin-Meadow, 2009), and it is even possible to predict words that will enter a child's lexicon based on their use of gestures at younger ages (Iverson & Goldin-Meadow, 2005). Deficits in gesture use have been proposed to contribute to poorer communication skills and pragmatic functioning in children with ASD (Crais et al., 2009; Tager-Flusberg, Paul, & Lord, 2005). It is hypothesized that gestures enhance language development by allowing children to adopt a more active role in social interactions, even at young ages when verbal language skills may be minimal. According to social-pragmatic theory of language development, engagement in joint attention episodes with an adult is one of the primary ways that children learn language (e.g., Tomasello, 2000). Gesture is thought to facilitate and enhance engagement in these joint attention episodes because it provides a channel for back-and-forth communication between the adult and the child that can be accessed even before the child has acquired spoken words (Rowe & Goldin-Meadow, 2009). Considering the strong empirical and theoretical link between early gesture use and later language development, describing profiles of early gesture use in populations at elevated risk for language delays has implications for early identification and intervention. However, very few studies have investigated gesture use as an early indicator of communicative impairment in high-risk populations such as ASIBs and infants with FXS.

Gestures in Infants With ASD and Non-ASD ASIBs

Infants who are later diagnosed with ASD show reduced gesture frequency, limited range of gestures, and differences in gesture functions. For example, by 12 months of age, infants who are diagnosed with ASD later in development are less likely than typically developing infants and infants with non-ASD developmental delays to use joint attention gestures and behavior regulation gestures (Mitchell et al., 2006; Osterling & Dawson, 1994; Rozga et al., 2011; Watson et al., 2013). Furthermore, Colgan et al. (2006) found that 9- to 12-month-old infants later diagnosed with ASD exhibited a more limited variety of social interaction gestures than typically developing infants.

Although atypical gesture use has been consistently reported in ASIBs later diagnosed with ASD, information on gesture use in non-ASD ASIBs is limited. Rozga et al.

(2011) found no evidence of atypical gesture use in non-ASD ASIBs, whereas another study reported that non-ASD ASIBs used fewer overall gestures and lower rates of behavior regulation gestures than low-risk control infants (Yirmiya et al., 2006). A more recent study investigated developmental trajectories of parent-reported gesture use in non-ASD ASIBs, both those with language delay and those without (Iverson et al., 2018). Results suggest non-ASD ASIBs without language delay exhibit fewer "early gestures" (e.g., giving, pointing, reaching) than low-risk controls starting at 9 months of age. Non-ASD ASIBs with language delay exhibited fewer early gestures at 8 months of age but with steeper rate of growth, resulting in a similar profile as non-ASD ASIBs without language delay by 14 months of age. These findings suggest that gesture use may be delayed in some non-ASD ASIBs in infancy, but inconsistent approaches to gesture measurement are contributing to an unclear developmental profile. Delayed or atypical emergence of gesture use in non-ASD ASIBs may provide a marker of risk for broader developmental difficulties, providing opportunity for early intervention and optimized outcomes. Thus, understanding the early communication profiles of ASIBs who do not go on to develop ASD is of critical importance.

Gestures in Infants With FXS

Although developmental delays are detected in infants with FXS as young as 9–12 months of age (Mirrett, Bailey, Roberts, & Hatton, 2004; Roberts, McCary, Shinkareva, & Bailey, 2016), there is a paucity of research on early communication in infants with FXS. One study by Marschik et al. (2014) suggested that 12-month-old infants with FXS exhibit a limited range of early communication skills, including deficits in eye contact, facial expressions, prelinguistic vocalizations (e.g., babbling), and gestures, but it is difficult to draw definitive conclusions from this study given the small sample size ($n = 7$) and lack of a control group. In another report, Hahn, Brady, McCary, Rague, and Roberts (2017) found that 7- to 14-month-old infants with FXS exhibited social-communicative behaviors, including gestures, that were less advanced than those exhibited by ASIBs and low-risk controls. The functions of the communicative acts also differed across groups, with the infants with FXS using communication for the purposes of behavior regulation and joint attention less often than the infant ASIBs and low-risk controls. However, the goal of this particular study was to capture the most complex communicative act exhibited by the infant in a given social interaction, and therefore, gestural acts were only coded if they represented the highest level of communication exhibited by the infant during that social press. Thus, the full range of gestural communicative acts exhibited by infants with FXS has yet to be described in the extant literature.

Profiling early gesture use is particularly important in the study of FXS because, given the significant language delays experienced by this group, young children with FXS may continue to use gestures as their primary mode of communication well into toddlerhood. In a descriptive study of 14- to 41-month-old children with FXS, Flenthrope

and Brady (2010) found that over half (65%) of the children's communication consisted of gestures, as opposed to vocalizations. Interestingly, in the subset of children with FXS who exhibited high levels of ASD symptoms, increased gesture use was correlated with poorer language skills at a 2-year follow-up. This suggests that children with FXS and elevated ASD symptoms may get "stuck" on gestures and fail to develop more advanced forms of communication. ASD symptoms have also been found to relate to the gesture profile of older 9- to 22-year-olds with FXS; Lorang and Sterling (2017) found that elevated ASD symptoms in children with FXS were associated with a greater proportion of gestures used to regulate others' behavior rather than to engage in social interaction/joint attention communicative functions. Overall, the literature suggests a divergent pattern of gesture use in FXS that likely interfaces with ASD risk in this group, although the gesture profile in infancy is not well understood.

This Study

Early communicative development in non-ASD infant ASIBs and infants with FXS is poorly understood, despite the fact that both groups are at elevated risk for ASD and its broader phenotype, language disorders, and other developmental delays. This study compared the frequency and function (i.e., behavioral regulation, social interaction, and joint attention) of communicative gesture use across non-ASD infant ASIBs, infants with FXS, and low-risk control infants at 12 months of age. We hypothesized that non-ASD infant ASIBs and infants with FXS would display reduced overall gesture frequency compared to low-risk infants and reduced use of gestures for social interaction and joint attention purposes. Profiling the function of early gesture use in non-ASD infant ASIBs and infants with FXS would inform early identification and treatment efforts for these groups and thus optimize communication outcomes.

Method

Participants

This study includes a cross-sectional analysis of gesture use at 12 months of age in 23 infant ASIBs who did not meet diagnostic thresholds for ASD later in development (i.e., non-ASD ASIBs), 18 infants with FXS, and 21 low-risk control infants. Data were collected as part of a larger longitudinal study of the early emergence of ASD in infants with FXS, for which the infants were followed from 6 to 36 months, as previously described (e.g., Guy, Richards, Tonnsen, & Roberts, 2018). All participants spoke English as the primary language in the home and had visual and hearing acuity within the normal range, per parent report. Group-specific inclusionary criteria are described below; group descriptives are presented in Tables 1 and 2.

Non-ASD ASIBs

All ASIBs were required to have an older sibling with a clinical diagnosis of ASD and no coexisting neurological

or genetic conditions. Because we were interested in the gesture use of non-ASD ASIBs, ASIBs who received a clinical best estimate diagnosis of ASD at 24 months of age or older were excluded from this study. Clinical best estimate procedures involved review of data from developmental assessments and gold standard ASD diagnostic tools by an expert multidisciplinary team that included a licensed psychologist and speech-language pathologist and are described in more detail in Hogan et al. (2017). Of the non-ASD ASIBs participants, one exhibited global developmental delay, defined by scores of ≤ 1.5 SDs below the mean on two or more Mullen Scales of Early Learning (MSEL; Mullen, 1995) subscales, with at least one of those subscales being Visual Reception, Fine Motor, or Gross Motor. Five exhibited subthreshold social-communicative differences and/or restricted and repetitive behaviors consistent with the BAP, defined by the presence of two or more *Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition* features of ASD (American Psychiatric Association, 2013), as determined by the professionals on the clinical best estimate team. None of the ASIBs exhibited isolated language delay. The remaining infants in the non-ASD ASIBs group were determined to have typical outcomes according to the clinical best estimate procedures (i.e., did not receive a clinical best estimate diagnosis of global developmental delay, BAP, isolated language delay, or ASD). ASIBs with non-typical outcomes were included in the sample because they represent the range of non-ASD outcomes observed in ASIBs.

FXS

The diagnosis of FXS was confirmed via genetic testing, and infants with FXS who had known comorbid genetic syndromes were excluded. Consistent with the broader literature, nine (50%) of the infants with FXS had comorbid ASD at 24 months of age or older according to clinical best estimate diagnoses. Considering the small sample sizes in the ASD subgroups of FXS ($n_s = 9$ within each subgroup), we were unable to address questions related to ASD comorbidity in FXS in this study. We lacked the statistical power needed to conduct valid inferential statistics, and descriptive analysis of the subgroup data is not informative given that the subgroups differed on several confounding variables, including sex (100% of infants who later developed ASD were male, whereas only 56% of those who did not develop ASD were male).

Low-Risk Controls

Low-risk control infants were required to have no family or personal history of ASD, FXS, or any related genetic disorders and to have been born at full-term gestation. Low-risk controls were included if they scored no lower than 1.5 SDs below the mean on the Early Learning Composite of the MSEL (Mullen, 1995). Given our focus on communication outcomes, low-risk infants who scored lower than 1.5 SDs below the mean on the Receptive or Expressive Language scales of the MSEL were also excluded.

Table 1. Group characteristics.

Characteristic	Group			Test of group differences (<i>p</i>)
	FXS <i>n</i> = 18	ASIBs <i>n</i> = 23	Low-risk controls <i>n</i> = 21	
Age (months)				
<i>M</i> (<i>SD</i>)	12.57 (0.66)	12.64 (0.69)	12.32 (0.42)	.190
Range	11.51–14.33	11.67–14.33	11.77–13.38	
Sex ratio (M:F)	13:5	16:7	16:5	.553
Race (%)				
Black or African American	17	13	5	.096
White	56	83	86	
More than one race	28	4	5	
Unknown or not reported	0	0	5	
Ethnicity (%)				
Hispanic or Latino	11	4	5	.580
Not Hispanic or Latino	89	96	90	
Not reported	0	0	0	
Maternal education level (%)				
High school	11	9	10	.382
Some college	6	26	10	
Associate's	11	9	9	
Bachelor's	44	30	38	
Some graduate	17	4	0	
Graduate degree	11	22	33	
Household income (%)				
≤ \$25,000	20	14	10	.480
\$25,001–50,000	7	19	25	
\$50,001–75,000	33	24	35	
\$75,000–100,000	7	24	5	
> \$100,001	33	19		

Note. FXS = fragile X syndrome; ASIBs = siblings of children with autism spectrum disorder; M = male; F = female.

Procedure

Assessments took place in a university laboratory setting or the infant's home and were completed in 1 day. Two trained examiners led each assessment, and infants' caregivers were present during the evaluations. After a

brief warm-up period, the protocol typically began with a play-based temperament assessment, followed by administration of the Autism Observation Scale for Infants (AOSI; Bryson, Zwaigenbaum, McDermott, Rombough, & Brian, 2008) and then the MSEL. Informed consent was obtained

Table 2. Descriptive statistics: Developmental level and early behavioral indicators of autism spectrum disorder.

Variable	Group			Test of group differences (<i>p</i>)
	FXS <i>M</i> (<i>SD</i>) Range	ASIBs <i>M</i> (<i>SD</i>) Range	Low-risk controls <i>M</i> (<i>SD</i>) Range	
MSEL Early Learning Composite standard score	80.17 (18.91) ^a 50.00–115.00	100.47 (14.13) ^b 74.00–137.00	104.43 (9.36) ^b 87.00–119.00	< .001
MSEL Visual Reception <i>t</i> score	41.89 (10.89) ^a 23.00–60.00	51.27 (13.25) ^b 31.00–80.00	51.76 (10.33) ^b 28.00–74.00	.017
MSEL Expressive Language <i>t</i> score	33.00 (11.77) ^a 20.00–57.00	45.05 (9.60) ^b 30.00–66.00	47.41 (9.92) ^b 36.00–66.00	< .001
MSEL Receptive Language <i>t</i> score	37.78 (11.61) ^a 20.00–60.00	44.00 (9.36) ^b 28.00–60.00	48.67 (8.26) ^b 35.00–64.00	.004
AOSI total score	9.60 (5.15) ^a 1.00–18.00	5.68 (3.23) ^b 1.00–13.00	4.33 (3.43) ^b 1.00–12.00	< .001
AOSI number of markers	5.93 (2.94) ^a 1.00–10.00	3.96 (1.94) ^b 1.00–9.00	2.86 (2.15) ^b 1.00–8.00	.001

Note. Groups sharing the same superscript did not differ significantly ($p \geq .050$). FXS = fragile X syndrome; ASIBs = siblings of children with autism spectrum disorder; MSEL = Mullen Scales of Early Learning; AOSI = Autism Observation Scale for Infants.

from the infants' parents, and all procedures were conducted in compliance with the University of South Carolina's Institutional Review Board.

Nonverbal Developmental Level

The MSEL (Mullen, 1995) is a test of cognitive and motor abilities that is appropriate from birth through 68 months of age. It includes five subscales: Gross Motor, Visual Reception, Fine Motor, Receptive Language, and Expressive Language. *T* scores were calculated for each individual subscale and combined to reflect an Early Learning Composite. Internal reliability is .75–.83 for subscales and .91 for the Early Learning Composite (Mullen, 1995). The Visual Reception subscale *t* score was used as an estimate of nonverbal developmental level, consistent with prior work (e.g., Hahn et al., 2017).

ASD-Related Risk Behaviors

The AOSI (Bryson et al., 2008) is a semistructured, play-based observational measure designed to capture early ASD symptoms in infants 6–18 months old. The AOSI served as the sampling context from which gestures were coded and provided an indicator of ASD-related risk behaviors within the sample. The AOSI consists of 19 items capturing ASD-related behaviors, scored on a scale from 0 to 3, with a higher score indicating increasing levels of abnormal behaviors. To describe the early presentation of ASD risk behaviors in the sample, two scores were computed: (a) a total score, which is computed as the sum of all items, and (b) the total number of markers (i.e., total number of items scored > 0), which has been shown to be the most discriminating of a later diagnosis of ASD. The presence of seven or more markers is considered a risk indicator for later ASD in high-risk populations (Bryson et al., 2008; Zwaigenbaum et al., 2005). The AOSI was administered by research staff who obtained official administration and coding reliability with the developers of the measure.

Gesture Frequency and Function

Gestures were coded from video recordings of the AOSI, which provides a play-based social interaction sample that is about 20 min in length. Gestures were coded using the schema outlined by Watson et al. (2013), which evaluates communicative gestures for both frequency and function. A communicative gesture was identified as any behavior that (a) acted as a gesture (e.g., physical movement of the hands, arms, and/or head), (b) was directed toward another person, and (c) served a communicative function. Examples of common gestures include nodding, pointing, clapping, and waving “bye.” Evidence that a gesture was directed toward another person was based on the pairing of the gesture with vocalization, looking at another person's face, touching another person, or having a shared engagement (e.g., contingent response to another person's words or actions). Provided that the behavior acted as a gesture and was directed at another person, it was considered a communicative gestural

act, and the function of the gesture was then determined. If a gesture served to obtain a desired object, get an individual to carry out an action, or to stop an individual from doing something, it was coded as *behavior regulation*. Gestures that attracted another person's attention in order to initiate or sustain a social game or routine (e.g., “peek-a-boo,” “patty-cake”), provided comfort to another, teased, obtained permission, or showed off were coded as social interaction gestures. Gestures that served the function of joint attention were aimed to direct another person's attention to a shared object or event in order to look at or comment about an object or event, to provide information about an object or event, or to confirm or acknowledge shared attention to an object or event. The total number of gestures and the number of gestures within each function class were included as dependent variables in this study. The full coding scheme is available from the authors upon request.

Each sample was coded via the consensus of two trained raters who were blind to group membership. Both raters coded each sample independently, and consensus scores, obtained by the raters via discussion of discrepancies across independent codes, were used in the analysis. Prior to coding, the raters completed extensive training that included joint coding and discussion of approximately 30 training videos and establishing at least 80% agreement on three consecutive videos. The kappa reliability statistic for reliability prior to consensus was computed at .52 for behavior regulation gestures, .66 for social interaction gestures, and .59 for joint attention gestures. In general, kappa values of .21–.40 indicate “fair” reliability, .41–.60 indicate “moderate” reliability, and .61–.80 indicate “substantial” reliability (Landis & Koch, 1977). Therefore, interrater reliability prior to consensus was “moderate” to “substantial” in magnitude, which is consistent with the reliability estimates achieved in prior studies employing this gesture coding scale (e.g., Watson et al., 2013). Low frequencies of occurrence can negatively impact kappa statistics (Hallgren, 2012), which may account for the relatively lower kappa values observed within the behavior regulation category.

Statistical Analysis

First, the mean number of gestures within each of the communicative function categories was examined descriptively across the groups. Examination of the raw data showed a zero-inflated distribution with each of the gesture variables exhibiting positive skew and overdispersion (i.e., the variance exceeded the sample mean). Thus, group differences in gesture use were tested using negative binomial regression models. Similar to the Poisson model, negative binomial models are appropriate for modeling count data exhibiting significant positive skew; however, unlike the Poisson model, the negative binomial model also includes an additional parameter, alpha, that accounts for overdispersion (White & Bennetts, 1996). Pearson chi-square goodness-of-fit statistics were examined for each model to confirm model fit. An exposure variable, which is used in count

models to adjust for individual differences in the length of observation, was included to capture variability in the total AOSI administration time. AOSI administration time did not differ across groups ($p = .956$). Covariates included biological sex, nonverbal developmental level, and maternal education level indexed as a three-level categorical variable reflecting education at the high school, bachelor's, or graduate level. Separate models were fit for the total number of gestures, as well as for the number of behavior regulation, social interaction, and joint attention gestures. A priori contrasts were employed to test differences in gesture use between the ASIBs and FXS groups versus low-risk controls, as well as differences between the ASIBs and FXS groups. Cohen's d effect sizes were calculated for all group contrasts, with d s of 0.20, 0.50, and 0.80 reflecting small, medium, and large effects, respectively (J. Cohen, 1988). Analyses were performed with SAS 9.4 using PROC GENMOD. To evaluate the relationship between gesture use, language ability, and ASD-related risk behaviors in infancy, partial Spearman correlations, controlling for nonverbal developmental level, were examined between the gesture variables, the MSEL Language subscales, and the AOSI total score.

Results

Descriptive Statistics

Table 3 reports the raw gesture data by group. Figure 1 illustrates the proportion of gestures serving each communicative function across the groups.

Group Differences in the Total Number of Gestures

Sex was a statistically significant predictor of overall gesture use, with females exhibiting a higher number of gestures, $\chi^2(1) = 19.31, p < .001$. Nonverbal developmental level and maternal education level did not contribute significantly to the model: $\chi^2(1) = 2.07, p = .150$ and $\chi^2(2) = 4.24, p = .120$, respectively. Planned group contrasts indicated that the FXS group used significantly fewer gestures than the low-risk controls ($p = .003, d = 0.87$) but did not differ significantly from the ASIBs ($p = .145, d = 0.43$). The ASIBs and low-risk controls did not differ significantly on the number of overall gestures used, although a medium effect size ($p = .070, d = 0.58$) was observed for fewer gestures in the ASIBs group. Group differences are illustrated in Figure 2.

Group Differences in the Number of Gestures by Function

Behavior Regulation Gestures

Sex and maternal education did not have a significant effect on the number of behavior regulation gestures used: $\chi^2(1) = 2.11, p = .146$ and $\chi^2(2) = 8.74, p = .120$, respectively. The effect of nonverbal developmental level was also non-significant, $\chi^2(1) = 1.14, p = .285$. Planned group contrasts indicated that the FXS group had significantly fewer behavior regulation gestures than the ASIB group ($p = .043, d = 1.02$). The FXS group did not differ from the low-risk controls, although Cohen's d indicated a medium effect size for group differences ($p = .263, d = 0.63$). The ASIBs and low-risk controls exhibited a similar number of behavior regulation gestures ($p = .296, d = 0.41$; see Figure 3).

Social Interaction Gestures

Sex was not significantly associated with use of social interaction gestures, $\chi^2(1) = 3.07, p = .080$. Nonverbal developmental level and maternal education did not contribute significantly to the model: $\chi^2(1) = 1.46, p = .227$ and $\chi^2(2) = 2.59, p = .274$, respectively. Both the FXS and ASIBs groups exhibited significantly fewer social interaction gestures than the low-risk controls, with large effect sizes for group differences ($p = .021, d = 1.14$ and $p = .018, d = 1.28$, respectively). The FXS and ASIBs groups did not differ from each other in the number of social interaction gestures ($p = .896, d = 0.27$). Group differences are illustrated in Figure 3.

Joint Attention Gestures

Sex was a statistically significant predictor of the number of joint attention gestures used, with females exhibiting more joint attention gestures, $\chi^2(1) = 20.28, p < .001$. The effects of nonverbal developmental level and maternal education level were not significant: $\chi^2(1) = 0.66, p = .418$ and $\chi^2(2) = 1.09, p = .580$. Planned group contrasts indicated no significant group differences between the FXS and low-risk groups ($p = .186, d = 0.53$) or the FXS and ASIBs groups ($p = .159, d = 0.64$), although the Cohen's d effect sizes for both contrasts were consistent with a medium effect size. The ASIBs and low-risk groups did not differ significantly ($p = .954, d = 0.01$; see Figure 3).

Table 3. Raw gesture data by group.

Group	Gesture index							
	Behavior regulation		Social interaction		Joint attention		Total gestures	
	<i>M</i> (<i>SD</i>)	Range	<i>M</i> (<i>SD</i>)	Range	<i>M</i> (<i>SD</i>)	Range	<i>M</i> (<i>SD</i>)	Range
FXS	0.6 (0.9)	0–3.0	3.0 (2.9)	0–10	1.6 (2.4)	0–7.0	5.2 (3.6)	1.0–13.0
ASIB	1.0 (1.1)	0–3.0	3.1 (1.2)	1–12.0	2.7 (2.9)	0–10.0	6.9 (5.3)	0–19.0
Low risk	0.8 (1.0)	0–3.0	6.0 (4.7)	0–15.0	3.0 (5.4)	0–25.0	9.8 (9.5)	1–40.0

Note. FXS = fragile X syndrome; ASIB = sibling of children with autism spectrum disorder.

Figure 1. Proportion of gestures used for each communicative function across groups. The numbers within the bars represent the mean number of gestures used within each communication function per group. ASIB = sibling of children with autism spectrum disorder; FXS = fragile X syndrome.

Relationship Between Gesture Use, Language Ability, and ASD-Related Risk Behaviors

Table 4 presents partial correlations between the gesture variables, the AOSI total score, and the MSEL Language subscales across participants, controlling for nonverbal developmental level. Within-group correlations were not conducted due to concerns regarding low statistical power related to the small sample sizes within the individual groups. Correlations within the full sample indicated that, for all gesture categories except behavior regulation, higher frequency of gesturing was significantly associated with fewer ASD-related risk behaviors (all $r_s > -.30$, all $p_s < .027$) and better expressive language skills (all $r_s > .27$, all $p_s < .047$). No significant correlations were observed between the gesture variables and receptive language (see Table 4).

Discussion

This study investigated the frequency and functions of gestural communication in 12-month-old non-ASD infant ASIBs and infants with FXS who are at high risk for

Figure 2. Total number of gestures by group. Model-predicted values are depicted. Groups sharing the same superscript did not differ significantly ($p \geq .050$). FXS = fragile X syndrome; ASIB = sibling of children with autism spectrum disorder.

Figure 3. Number of gestures within each communicative function by group. Model-predicted values are depicted. Within each communicative function, groups sharing the same superscript did not differ significantly ($p \geq .050$). FXS = fragile X syndrome; ASIB = sibling of children with autism spectrum disorder.

developing ASD and communication disorders. Findings indicated that, after accounting for nonverbal developmental level, infants with FXS used gestures less frequently than low-risk infants, whereas non-ASD ASIBs did not differ from either the infants with FXS or the low-risk controls in the overall frequency of gesturing. The analysis of communicative function provided a more nuanced picture, where both non-ASD ASIBs and infants with FXS used gestures for social interaction less frequently than low-risk infants. Neither infants with FXS nor infant ASIBs differed significantly from low-risk controls in the frequency of behavior regulation gestures, although examination of group means suggests a potential weakness in the use of behavior regulation gestures in the FXS group. No group differences were detected in the frequency of joint attention gestures. Thus, findings support the presence of divergent communication features in non-ASD ASIBs and infants with FXS that are detectable as early as 12 months of age. This study builds upon the limited knowledge of early communication phenotypes in infants with FXS and infant ASIBs who do not go on to meet diagnostic criteria for ASD and has implications for the clinical evaluation and treatment of infants who are at high risk for ASD and communication disorders.

Gesture Frequency

This study examined both gesture frequency and function. Gesture frequency is a global measure capturing the frequency of any communicative gestures exhibited by the child, without considering the function of the communicative act. In clinical practice, observation of gesture frequency may represent a more accessible index than gesture function; while many clinicians will feel comfortable marking when a gesture has occurred, it takes considerably more training to determine the communicative function of the gesture.

Table 4. Partial correlations between gesture use, receptive and expressive language, and autism spectrum disorder risk scores across all participants, controlling for nonverbal developmental level.

Gesture variable	MSEL Receptive Language <i>t</i> score	MSEL Expressive Language <i>t</i> score	AOSI total score
Overall frequency	.21	.42*	-.39*
Behavioral regulation	-.05	.25	-.15
Social interaction	.13	.27*	-.30*
Joint attention	.25	.41*	-.41*

Note. MSEL = Mullen Scales of Early Learning; AOSI= Autism Observation Scale for Infants.

* $p < .050$.

Yet, our results indicate that reliance on gesture frequency provides an incomplete picture of early gestural development. When considering the overall frequency of gestures, 12-month-old infants with FXS displayed a delayed communicative profile relative to low-risk infants that was of a large effect size, even after accounting for nonverbal developmental level. However, gesture frequency was not sensitive enough to differentiate the non-ASD ASIBs from either the FXS or low-risk groups. Further examination of gesture functions showed that non-ASD ASIBs nonetheless exhibited an atypical profile of gesture use characterized by reduced use gestures for social interaction communicative functions, as discussed below. Thus, clinicians who rely on information about the overall frequency of gestures but fail to consider gesture function may overlook important early communicative differences in infants who are at risk for ASD and communication impairment.

Behavioral Regulation Gestures

Consistent with the findings of Hahn et al. (2017), our findings suggest that infants with FXS were less advanced in their ability to use gestures to influence others' behavior. Significantly fewer behavior regulation gestures were observed in infants with FXS relative to ASIBs, with a medium effect size. The FXS group also showed a lower mean frequency of behavior regulation gestures relative to the low-risk controls that was not statistically significant, although the medium effect size for group differences suggests that we may have been underpowered to detect this effect. Notably, delayed nonverbal developmental level did not account for less frequent behavior regulation gesture use in infants with FXS.

Follow-up studies are needed to determine the correlates of behavior regulation gesture use in infants with FXS; it is possible that this profile reflects early delays specifically within the domain of communication, independent of nonverbal developmental level. Another possibility is that reduced use of behavioral regulation gestures reflects heightened ASD risk, as other studies have found reduced behavior regulation gestures in infants with idiopathic ASD (Rozga et al., 2011; Watson et al., 2013). Yet, evidence from a study of gestures in older children with FXS suggests the opposite, where increased ASD symptoms have been linked with a greater proportion of gestures used for the purpose of

behavior regulation (Lorang & Sterling, 2017). In our own sample, behavior regulation was the only gesture variable unrelated to the severity of ASD-related risk behaviors exhibited by the infants. This suggests that ASD risk did not track closely with behavior regulation gestures use in this sample of 12-month-old infants. Though findings are preliminary given that we lacked the sample sizes necessary to evaluate the presence of group-specific patterns, they encourage the future examination of the association between ASD symptoms/comorbidity and gesture use in infants with FXS. Such knowledge will refine our understanding of the interface between ASD-related behaviors and communication development in this group.

It is interesting to note that behavior regulation gestures occurred relatively infrequently across all groups of participants (ranging from zero to three instances). It is unclear how this frequency compares to prior research, given that no prior investigations have reported counts of behavior regulation gestures occurring during the AOSI or other similarly structured examiner-child interaction contexts, and gesture frequency likely varies according to both child-related characteristics and contextual factors, such as familiarity with communication partner, locus of control, level of structure, and the types and novelty of materials used. The establishment of normative data on the expected rate of gestural communication will be important in future research, as this information would greatly assist clinicians in recognizing when gesture use deviates from the norm.

Social Interaction Gestures

Reduced frequency of social interaction gestures was a striking feature in both non-ASD infant ASIBs and infants with FXS, with large effect sizes separating these groups from low-risk controls. This is notable because the infant ASIBs in our study, who did not later meet criteria for ASD, nonetheless exhibited gestural communication features that deviated from low-risk infants at 12 months of age. This pattern of reduced used of social interaction gestures may be reflective of risk for other nontypical developmental outcomes in this group, such as the BAP, which was observed in about 20% of the present ASIB sample. The observed association between social interaction gesture use and variation in language ability and ASD-related behaviors in the infants provides preliminary support for this hypothesis.

Although we detected robust differences between the non-ASD ASIBs and low-risk groups in social interaction gesture use, some prior research has not found social interaction gestures to be impaired in infants with ASD, who would be expected to show more prominent communication deficits than non-ASD ASIBs (e.g., Watson et al., 2013). The AOSI sampling context in this study likely enhanced our ability to evaluate social interaction gesture use. The AOSI includes two social interaction games, rolling a ball back and forth and a “peek-a-boo” press, which ensures that every infant is given opportunities to use gestures to initiate/sustain interaction within a familiar social routine. In fact, 55% of all social interaction gestures observed in this study occurred during the “peek-a-boo” press of the AOSI (data not presented). This is consistent with the findings of Hahn et al. (2017), who found that infants with FXS used more sophisticated social communication behaviors during the “peek-a-boo” press of the AOSI than during any of the other AOSI contexts. These convergent findings suggest that incorporating common social routines into clinical interactions (e.g., teasing “I’m going to get you” tickle games, “this little piggy went to the market,” patty-cake) can provide a rich social context for evaluating and targeting social-interactive communicative behaviors. Indeed, many naturalistic developmental behavioral interventions for children with ASD explicitly incorporate social play routines as a context that offers shared control and fosters child-initiated teaching episodes by increasing motivation and providing natural positive reinforcement (Schreibman et al., 2015).

Joint Attention Gestures

Although joint attention is a known area of weakness in children with ASD, we did not detect differences in the number of joint attention gestures used by the FXS, ASIBs, or low-risk infants. Our relatively low sample sizes, coupled with the large variability in the frequency of joint attention gestures exhibited by the low-risk controls, may have limited power to detect statistical effects. It is notable that infants with FXS, on average, used lower mean levels of joint attention gestures than both the ASIBs and low-risk group, with medium effect sizes. In contrast, comparable mean levels of joint attention gestures were observed across ASIBs and low-risk controls, with a small effect size suggesting negligible group differences. Considering that joint attention deficits are well documented in ASD (e.g., Charman et al., 1997; Mundy, Sigman, & Kasari, 1994), the similarity across the ASIBs and low-risk groups in joint attention gesture use is somewhat surprising, although the finding is consistent with several other reports focused on ASIBs. For example, Cassel et al. (2007) did not detect group differences in joint attention gestures between 12-month-old low-risk infants and ASIBs whose ASD outcomes were unknown. Rozga et al. (2011) was also unable to detect group differences in joint attention gestures between 12-month-old ASIBs later diagnosed with ASD and low-risk infants.

It is possible that 12 months of age may be too early to detect delays in joint attention gesture development, as

joint attention gestures are still emerging in this developmental period, with low overall rates of joint attention gestures observed across all 12-month-old infants (e.g., Watson et al., 2013). Although the overall frequency of joint attention gestures did not distinguish the groups, joint attention gesture use was nonetheless associated with the severity of ASD-related risk behaviors and expressive language skills of the infants, which is consistent with the broader literature demonstrating close ties between joint attention skills and language/communication ability in both typical and clinical groups (e.g., Charman, 2003; Morales et al., 2000). In summary, this study adds to the extant literature suggesting that joint attention gestures do not reliably distinguish non-ASD ASIBs from low-risk controls during infancy. According to our data, reduced use of social interaction gestures during familiar social routines may provide a more reliable indicator of risk at this age.

Clinical Implications

This study has a number of clinical implications. First, our findings support gesture use as an informative marker than can be used, in addition to spoken language measures, to obtain a comprehensive picture of communicative abilities in young children. Likewise, clinicians evaluating and targeting gestures should consider not only the frequency at which gestures are occurring but also the communicative function of the gestures. Simple frequency counts were able to differentiate infants with FXS from low-risk infants but were not sufficient for capturing the atypical communication patterns in the non-ASD ASIBs or the more nuanced areas of weakness in the infants with FXS. Clinicians who are cognizant of the functions of communication will be better able to evaluate and monitor the communication strengths and weaknesses of young children who are at risk for ASD and/or language and communication difficulties. Speech-language pathologists have a variety of tools at their disposal to evaluate the quality of gestural communication, including standard behavioral observation methods such as the approach used in this study, as well as more structured assessments that will yield information on the function of gestural and other nonverbal communicative acts, such as the Communication Matrix (Rowland, 2004), the Communication and Symbolic Behavior Scales (Wetherby & Prizant, 2003), and the Inventory of Potential Communicative Acts (Sigafoos, Arthur-Kelly, & Butterfield, 2006).

It is clear that speech-language pathologists should consider gestural communication as an intervention target, given that this foundational skill is a strong predictor of later language development, both in typical development (e.g., Iverson & Goldin-Meadow, 2005; Orr, 2018) and in children with developmental disorders (e.g., Manwaring, Mead, Swineford, & Thurm, 2017; Özçalışkan, Adamson, Dimitrova, & Baumann, 2017). The results of this study may have some implications for targeting gestures. First, joint attention gesture use did not distinguish the at-risk infants in this study from the low-risk infants, perhaps suggesting that 12 months may be too early to detect atypical

joint attention gesture use. In contrast, gestures for the purpose of social interaction were strikingly reduced in both the infants with FXS and the non-ASD ASIBs. Therefore, instead of focusing on joint attention gestures, clinicians working with this age group might instead focus on social interaction gestures as a useful risk marker and potential treatment target. Second, it is notable that over half of the observed social interaction gestures occurred during the familiar social routine of “peek-a-boo,” suggesting that social routines may provide a rich context for promoting high-quality communicative behaviors. Clinicians might capitalize on the communicative strengths that occur during familiar social routines to help boost the use of these skills in other contexts. For example, clinicians may work to gradually encourage social interaction gestures across a wider variety of contexts, perhaps by first teaching skills during structured social games that may be less well practiced (e.g., “London Bridge,” “I’m a Little Teapot,” “Teddy Bear Teddy Bear Turn Around”) and eventually moving into nonrehearsed social routines that arise spontaneously through playful interaction (e.g., a back-and-forth game of jumping and pausing or a silly back-and-forth game of accidentally putting shoes on hands and mittens on feet—in other words, any “routine” that is composed of predictable back-and-forth social exchanges where both the clinician and the child contribute turns). The use of “teachable moments” that evolve from social routines is consistent with the approach used in many naturalistic developmental behavioral interventions, which capitalize on social routines as an ideal context for encouraging child-led teaching opportunities and increasing motivation and joint engagement (Schreibman et al., 2015). Examples of naturalistic developmental behavioral intervention approaches that include an explicit focus on gestures are Reciprocal Imitation Training (Ingersoll & Lalonde, 2010) and the Early Start Denver Model (Dawson et al., 2010).

Limitations and Future Directions

This study has a number of strengths, such as direct observation measures of gesture ability and the use of a semi-structured play-based interaction that ensured that gestures were evaluated from a similar communication context across all infants. Another noteworthy strength is the study of gesture use in infancy. FXS is not generally diagnosed until 3.5 years of age (Bailey, Raspa, Bishop, & Holiday, 2009), making the study of infants with this genetic condition a significant challenge. This study adds to the few existing reports describing early communication skills of infants with FXS. Additionally, our inclusion of ASIBs allowed us to contrast the early gesture profile of FXS with that of other populations who are at increased risk for ASD and related communication disorders, thus shedding light on potential syndrome-specific profiles. However, as is true of many studies involving special populations, we were limited by small sample sizes. In particular, the small number of infants with FXS prevented us from exploring potentially informative questions, such as questions regarding the impact

of ASD comorbidity on gesture use in the FXS group. Another interesting direction would be to further tease apart the relationship between early gesture use and risk for specific nontypical developmental outcomes in the ASIBs. Although we know that around 25% of the present ASIBs sample had nontypical developmental outcomes (e.g., social communication features of the BAP or global developmental delay), we lack the sample sizes necessary to examine gesture use as a differential prognostic indicator in this group.

This report focused specifically on gestures as characterized within the taxonomy of Bruner’s (1981) communicative functions. Future studies may include analysis of other properties of gestures, such as the gesture type. Investigations into gesture type may provide a deeper understanding of the range, flexibility, and sophistication of communicative forms observed across at-risk groups. Gestures can turn into a form of augmentative and alternative communication for children with complex communication needs, and therefore, the profile of both gesture function and form are considerations when designing a treatment plan for a particular child. Another future direction includes the study of gesture use during naturalistic caregiver–child interaction. Stronger gestures skills may be observed in this context, given that caregivers of children with developmental disabilities may compensate for their child’s limited responsiveness by providing increased scaffolding (e.g., Baranek, 1999; Watson et al., 2013). Better understanding of the influence of context/communication partner/level of structure on gesture use will inform the consideration of these factors in assessment and treatment.

Conclusion

By profiling communicative gesture use in 12-month-old infants with FXS and ASIBs who do not later meet diagnostic criteria for ASD, this study sheds new light on early-emerging communicative difficulties that may represent targets for intervention. Our findings suggest that the overall frequency of gestures is reduced in infants with FXS, even after controlling for the influence of nonverbal cognitive delay. Analysis of gesture function revealed more nuanced information on communication patterns that diverged across the high- and low-risk groups. Compared to low-risk controls, infants with FXS and non-ASD ASIBs displayed fewer gestures for the purpose of social interaction at 12 months of age. The use of behavioral regulation communication functions was also reduced in the infants with FXS. Significant associations between gesture use and variation in language ability and ASD-related risk behaviors in infants supports the notion that impaired gesture use may represent a risk marker for later spoken language difficulties and the BAP. These findings provide important insight into early developmental profiles relevant to FXS, ASD, and BAP and highlight the need to more closely examine gestures as a mechanism to aid in early identification and intervention in future work.

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