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Assessing Joint Attention Responding and Initiation in Children with Angelman Syndrome

Jane Summers*,† and Julie Impey‡

*McMaster Children's Hospital, Hamilton Health Sciences, Hamilton, Ontario Canada; †McMaster University, Hamilton, Ontario Canada; ‡Deer Lodge Centre, Winnipeg, Manitoba Canada

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Background Angelman syndrome (AS) is a rare neurogenetic disorder that overlaps with autism and is associated with severe cognitive, speech and motor impairments. Communication and social deficits are found among children with AS, both of which may be linked to impairments in joint attention.

Methods The participants in this study were four children with a genetically confirmed diagnosis of AS, aged 5–10. Children's joint attention responding and initiation were measured using a modified version of a structured assessment protocol from MacDonald *et al.* (2006).

Results Children's ability to respond to joint attention bids was less impaired than their ability to initiate joint attention.

Conclusions Differences in children's response patterns are discussed in relation to their characteristics on measures of cognitive, adaptive and language functioning as well as on a measure of autistic symptomatology. A better understanding of the nature of joint attention deficits in children with AS may assist with developing remedial approaches to improve their communication and social skills.

Keywords: Angelman syndrome, behavioural assessment, joint attention

Introduction

Angelman syndrome (AS) is a neurogenetic disorder that is caused by the lack of expression of the maternal copy of the UBE-3A gene on chromosome 15 (Kishino *et al.* 1997). AS is characterized by severe intellectual disability, seizure disorder, motor dysfunction, absent or minimal expressive speech and happy demeanour with frequent bouts of laughter (Angelman 1965). The prevalence of AS is estimated to be between 1/10 000 and 1/20 000 births (Clayton-Smith & Pembrey 1992; Petersen *et al.* 1995). There are four main classes of AS (deletion of 15q11-q13, imprinting defects, paternal uniparental disomy and mutations in the UBE3A gene), accounting for up to 90% of cases (Kishino *et al.* 1997; Jiang *et al.* 1999). The remainder of cases are diagnosed on the basis of clinical features (Williams *et al.* 2006). Children with the deletion of 15q11-q13 tend to have the most severe neurodevelopmental deficits and most restricted range of skills (Varela *et al.* 2004).

There is a pronounced gap between expressive and receptive language abilities in children with AS (Gentile *et al.* 2010). Receptive language is often sufficient for them to comprehend simple instructions (Jolleff & Ryan 1993), whereas expressive language, when it does develop, is limited primarily to single words (Andersen *et al.* 2001). Owing to their lack of functional speech skills, children with AS may rely on augmentative and alternative communication (AAC) systems as their primary or supplementary means of communication. Few studies contain specific information about AAC usage by children with AS and are primarily descriptive in nature. The use of aided approaches such as photographs, picture communication symbols and voice output devices has been reported (Summers *et al.* 1995; Alvares & Downing 1998; Andersen *et al.* 2001; Calculator 2002; Didden *et al.* 2009), but the relative effectiveness of such approaches has not been systematically evaluated. It appears that children are most often able to indicate their preferences and needs through physical

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means such as reaching, pointing, manipulating other people's hands and head nods and shakes (Jolleff & Ryan 1993; Penner *et al.* 1993). Another finding is that gestures are used more commonly than manual signs (Jolleff & Ryan 1993; Penner *et al.* 1993). As with words, signs are often approximations and are difficult to understand because of children's motor impairments (Alvares & Downing 1998; Calculator 2002; Jolleff *et al.* 2006).

Duker *et al.* (2002) used a rating scale to assess different forms of communicative behaviour (requesting/rejecting, labelling and imitating other people's gestures and words) among children with AS, Down syndrome and pervasive developmental disorder. The mode of children's expressive communication differed significantly as speech was used by more than 75% of children in the contrast groups but not by any of the children with AS. Although there were no group differences in relation to the functions of requesting and labelling, children with AS were much less likely to imitate communicative behaviour than children in the two other groups. In a large-scale study involving 109 participants with AS that employed the same rating scale, Didden *et al.* (2004) found that children with AS engaged in communicative behaviour (requesting) primarily to have their wants and needs met, whereas age- and ability-matched children used more advanced forms of communication (i.e. labelling and imitating other people's gestures or words) that serve to bring about and maintain social interactions. Among the children with AS, greater impairments in communicative behaviour were associated with lower levels of intellectual functioning as well as with the presence of a seizure disorder and the use of anticonvulsant medication. One hypothesis to emerge from this study is that seizures and anticonvulsant medication negatively impact the development of communication skills in children with AS.

It appears likely that prelinguistic factors play a role in children's communication impairments (Jolleff & Ryan 1993). Alvares & Downing (1998) conducted a parent survey of expressive communication skills among children with AS, reporting that only about 50% of the children with AS in their sample maintained eye contact during a communicative exchange and only 20% intentionally gained their partner's attention prior to signing. Penner *et al.* (1993) found that only one of seven adults with AS they assessed using a pre-language communication measure was able to jointly attend and act on objects with another person and to participate in a turn-taking exchange. This individual was also able to point and use other types of gestures to communicate. Jolleff & Ryan (1993) reported the majority of children they assessed using a preverbal communication schedule rarely imitated motor actions and failed to use gestures as a means to communicate. The investigators also noted that children frequently lacked referential eye gaze. Imitation plays an important role in early development as it provides a means for infants and children to learn new skills as well as to engage in social interactions (Ingersoll 2008). Imitation is also linked to joint attention, which refers to the ability to coordinate visual attention with a social partner (Mundy & Newell 2007). Joint attention refers to separate but inter-related behaviours that emerge in infancy and serve as a foundation for social-cognitive development (Mundy & Gomes 1998; Mundy & Newell 2007). Two types of joint attention have been identified which may involve different neural systems. Responding to joint attention involves the ability to follow the gaze and gestures of another person to share a common social point of visual reference, whereas initiation of joint attention involves spontaneously using one's own gaze and gestures to direct the attention of another person to objects or events as well as themselves (Mundy & Newell 2007; Mundy *et al.* 2009).

Outside of the infant development field, the relationship between joint attention and social and communication skills has perhaps been most extensively studied in young children with autism spectrum disorders (ASD) (e.g. Mundy *et al.* 1986; Charman 2003). Significant impairments in symbolic communication, imitation and joint attention skills have been identified in this group of children, all of which are associated with the development of language (Toth *et al.* 2006). There is evidence that children with autism are less impaired in responding to joint attention than initiation of joint attention (MacDonald *et al.* 2006; Mundy & Newell 2007). In fact, impairments in the initiation of joint attention are considered a core deficit in ASD.

There is emerging evidence of an overlap between AS and ASD, particularly in regard to impairments in children's social communication, imitation and joint attention abilities (Steffenburg *et al.* 1996; Peters *et al.* 2004; Trillingsgaard & Ostergaard 2004). Some of the possible reasons for this association include severe-to-profound intellectual disability, presence of epilepsy and abnormalities of the AS chromosomal region (15q11-q13) (Bonati *et al.* 2007).

Steffenburg *et al.* (1996) found that children with AS who met the criteria for a diagnosis of autism failed to spontaneously seek others to share enjoyment or inter-

ests, a deficit that relates to impairments in the initiation of joint attention. Trillingsgaard & Ostergaard (2004) reported that children with AS and comorbid autism (based on the Autism Diagnostic Observation Schedule), much like their counterparts with a diagnosis of autism only, showed almost non-existent use of communicative gestures in an instrumental way and a lack of joint attention behaviours. There was an inverse relationship between children's mental age scores (particularly their receptive language scores) on a measure of cognitive ability (Mullen Scales of Early Learning) and the severity of their autism symptoms. Peters *et al.* (2004) found that children with comorbid AS and autism diagnosis demonstrated impairment in joint attention abilities as they did not display shared enjoyment in interactions with others and tended to direct their attention to objects, rather than objects and other people at the same time. These same children also showed slower rates of growth over a 12-month period on measures of cognitive, language and adaptive functioning than their peers with AS who did not receive a diagnosis of autism. It is possible that low levels of cognitive and language ability may directly underlie deficits in social communication functioning among children with AS.

The purpose of this study was to obtain detailed information regarding joint attention behaviours in four children with AS. As children with AS have more severely impaired expressive communication skills than would be expected on the basis of their level of intellectual functioning alone, attempts have been made to identify other factors that may have a bearing on this issue. Previous researchers have reported an overlap between AS and features of ASD, particularly in regard to deficits in children's social communication and joint attention abilities. In this study, children's joint attention responding and initiation were measured directly using a structured assessment protocol that was adapted from MacDonald *et al.* (2006). Individual differences in children's response patterns were explored in relation to their characteristics on measures of cognitive and language functioning and an assessment of autistic symptomatology.

Method

Participants

Four children with AS (three girls and one boy) participated in the study. The children were between the ages of 5.4 and 10.3 years and lived at home with both their parents. They had all undergone genetic testing to establish a diagnosis of AS and exhibited classic features of the syndrome including lack of speech, motor impairments, attention problems and happy demeanour. Three children had a maternal deletion of 15q11-q13, while the fourth child had a mutation of the maternal UBE-3A gene. The deletion positive children all had a seizure disorder that was being treated with anti-convulsant medication, while the child with the gene mutation did not. Three of the four children were integrated into classes with typically developing children and received individualized support from an Educational Assistant. The fourth child was placed in a segregated classroom with a low teacher-to-student ratio. The children were involved in a larger study to evaluate the effectiveness of approaches (discrete trial teaching or verbal behaviour) that were based on the scientific teaching principles of Applied Behaviour Analysis for improving functional skills in children with AS (Summers & Szatmari 2009) and had been participating in the intervention study from 1 to 3 years. The instructional curriculum was designed to improve children's skills in the areas of attending to people and objects, expressive communication (requesting preferred items) and motor imitation, among others. As part of this study, children underwent a developmental assessment by an independent psychologist. The Mullen Scales of Early Learning were used as a measure of their cognitive development (Mullen 1995). Their adaptive functioning was assessed using the Vineland Adaptive Behavior Scales, Interview Edition (Sparrow *et al.* 1984), and the Receptive and Expressive Emergent Language Scale, Second Edition (Bzock & League 1991), was used as a language measure. The last two measures are based on parental report, whereas the Mullen is based on direct observation of a child in a standardized testing situation. For each assessment measure, raw scores were transformed into age-equivalent scores. An assessment of autistic symptomatology using the ADOS-G Module 1 (Lord *et al.* 2002) was completed for descriptive rather than diagnostic purposes. Parents of all four children provided consent for their children to participate in an undergraduate thesis project to study joint attention. The study had received approval by the University Research Ethics Board. Characteristics of the participants are presented in Table 1.

Procedure

The assessment protocol that was used to measure children's joint attention response and initiation was adapted from the procedures used by MacDonald *et al.*

(2006). Two subtests were used to evaluate children's response to joint attention bids by the examiner, and three subtests were used to evaluate their ability to initiate joint attention bids. Assessments were made in the children's homes in a room that was used for their therapy sessions and was relatively free from visual and auditory distractions. During the assessments, the examiner and child were seated across from and facing each other at a table. Each child's therapist and one of his or her parents were present or nearby during the assessment. Sessions were videotaped to collect reliability data. The entire assessment took approximately 4–5 min to complete.

Joint attention responding

Children's joint attention responding was assessed using pictures and toys.

Response to pictures. The response to pictures subtest utilized a folder that contained six brightly coloured pictures of animals, one picture per page. The examiner opened the folder and laid it flat on the table, directly in front of and facing the child. The examiner attempted to establish eye contact with the child by calling his or her name. If the child did not look in the direction of the examiner's eyes within 2 or 3 s, the examiner made a sweeping motion with her fingers and brought them up to her eyes while saying 'Look at me.' Once the child made eye contact, the examiner directed her gaze to the first target picture in the folder. A maximum score of 3 was awarded if the child responded correctly by following the direction of the examiner's gaze shift to look towards the target picture within 5 s and then the trial ended. If the child did not make a correct response, the examiner attempted to re-establish eye contact by calling the child's name and making a sweeping motion by her eyes if needed. When the child was again looking in the direction of the examiner's eyes, she pointed at the picture with an extended index finger while shifting her gaze to it at the same time. A correct response to the examiner's point and gaze shift was awarded a score of 2 and the trial ended. If the child still did not look towards the target picture within 5 s of a point plus a gaze shift, the examiner followed the steps outlined earlier to obtain eye contact and said 'Look' while pointing at the picture with an extended index finger and shifting her gaze to it at the same time. A correct

Table 1 Characteristics of participants

Measure	*Child 1*	*Child 2*	*Child 3*	*Child 4*
Gender and chronological age (years-months)	Female	Male	Female	Female
	10-1	5-4	6-3	10-3
Molecular subtype	Del +	Del +	Del +	UBE3A mutation
Presence of seizures	Yes	Yes	Yes	No
Length of time in intervention study	3.5 years	2.5 years	2.5 years	1 year
Mullen Scales of Early Learning (age-equivalent scores in years-months)				
Expressive language	0–5	0–4	0–6	0–7
Receptive language	1–5	1–1	1–3	2–3
Visual reception	1–8	0–11	1–4	3–0
Vineland Adaptive Behavior Scales (age-equivalent scores in years-months)				
Communication	1–4	0–9	1–0	1–4
Socialization	1–4	0–10	1–1	1–7
Receptive-Expressive Emergent Language Scale – Second Edition (age-equivalent scores in years-months)				
Expressive language	0–5	0–3	0–5	0–6
Receptive language	1–0	0–10	0–11	2–6
Autism Diagnostic Observation Scale – generic (module 1)				
Communication	6	5	3	0
Reciprocal social interaction	1	10	4	0
Total score	7	15	7	0
ADOS classification	No ASD[1]	Autism	ASD	No ASD

[1]Child did not meet cut-off for reciprocal social interaction on the ADOS-G so is not classified as falling within the autism spectrum.
ASD, autism spectrum disorders.

response to the examiner's verbal directive, point and gaze shift was awarded a score of 1 and the trial ended. If the child did not look at the target picture when all three cues were presented, the trial ended and a score of 0 was assigned. This procedure was repeated for the third and fifth pictures in the folder. For the second, fourth and sixth pictures in the folder, the child was permitted to look at the pictures while the examiner informally interacted with him or her.

Response to objects. For the response to objects subtest, the identical sequence that was outlined under response to pictures was repeated. Three brightly coloured toys (the most highly ranked items that were previously identified in a forced-choice preference assessment) were placed at the child's eye level in visible locations around the room prior to the commencement of the session. A score of 3 was awarded if the child followed the direction of examiner's gaze shift to look towards the target object; a score of 2 was awarded if the child followed the direction of the examiner's point and eye gaze; a score of 1 was given if the child followed the examiner's verbal directive to look along with a point and shift in eye gaze; and a score of 0 was given if the child did not respond when all three cues were given.

Joint attention initiation subtests

Children's initiation of joint attention was assessed using two toys that were activated and a book for the child to look through. For the *bubble machine* subtest, a purple hippopotamus-shaped bubble machine was brought out from its hiding spot under the table and placed on top of the table, remaining visible but out of reach of the child. The examiner activated the bubble machine and observed the child's reaction for approximately 15 s. After 15 s had elapsed, the bubble machine was turned off and the child's reaction was observed for an additional 5 s. For the *toy car* subtest, a toy car that had been placed on the floor out of reach of the child was activated using a remote control. The examiner observed the child for 15 s while the car was moving and 5 s after the car was turned off. For the *book presentation* subtest, the examiner opened a book containing several pictures on each page and placed it on the table in front of the child (*Nubby Bunny*; McMullan 2003). The examiner allowed the child to look at the book, touch it and turn the pages for approximately 20 s.

For all five subtests, the examiner responded to the child's interest in the item with a brief verbal comment ('Look at that' or 'That's great'). A gaze shift was recorded if the child looked at the toy during the activation period or after the toy was turned off or looked at the book during the presentation period and then immediately looked at the examiner. The number of gaze shifts per subtest was scored. One gaze shift was recorded if the child looked from the object to the examiner, two gaze shifts were recorded if the child looked from the object to the examiner and back to the object again and so on. A new gaze shift was recorded if the sequence was interrupted and then started all over again. A gesture was recorded if the child pointed to or reached towards the toy or book or himself/herself while looking at the examiner or the toy/book. A vocalization was recorded if the child made a sound (which could include laughter) while looking at the toy/book or examiner during the activation or presentation period.

Interobserver agreement

Children's responding on the joint attention response and initiation subtests was recorded from videotapes by the examiner and an independent observer. Interobserver agreement data were obtained from the assessment sessions for all four children and were calculated by dividing the number of agreements by the number of agreements plus disagreements and multiplying the resulting number by 100 to yield a percentage score. Mean per cent agreement for the joint attention response subtests was 94% (96% for response to gaze shift and 92% for response to pointing). Mean per cent agreement for presence/absence of joint attention initiation behaviours was 93% (85% for child gaze shift, 100% for gestures and 95% for vocalizations) and for total number of gaze shifts was 85%.

Results

Children's individual results on the joint attention response subtests are presented in Figure 1. Scores for all six joint attention subtests were added together to yield a total composite score. For each subtest, the child was given a score of 3 for responding to the examiner's shift in eye gaze, a score of 2 for responding to shift in eye gaze with a point and a score of 1 for responding to shift in eye gaze when combined with a point and verbalization. A score of 18 was the maximum possible composite score for each child. Three of the four children received an identical score of 15, based upon their ability to follow the direction of the examiner's gaze shift to look towards an object or picture on 50% of the

Figure 1 Individual composite scores on joint attention responding subtests by type of examiner bid.

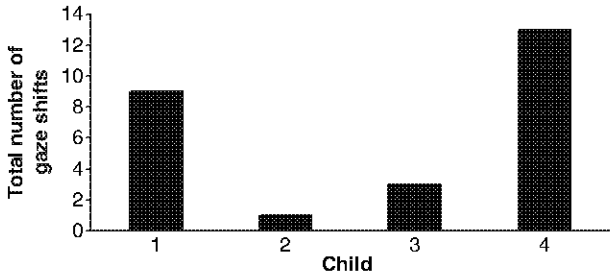

Figure 3 Total number of gaze shifts for each child during joint attention initiation subtests.

subtests and their responding to the examiner's gaze shift and point on the remaining subtests. By contrast, the fourth child (Child 2) received a score of 12 based on his responsiveness to the examiner's shift in eye gaze only when it was accompanied by a point.

Figure 2 shows children's individual results on the joint attention initiation subtests. Composite scores were based on the occurrence of the three different types of joint attention initiation behaviours (gaze shift, gesture and vocalization) during each of the three subtests, for a maximum score of 9. Two of the four children (Child 1 and Child 4) demonstrated all three types of joint attention initiation behaviours, while the other children demonstrated only two types of these behaviours. The use of gaze shift to initiate joint attention was used by all four children.

Figure 3 shows the total number of gaze shifts demonstrated by each of the children across all three joint attention initiation subtests. Scores ranged from 1 to 13, with two of the four children (Child 1 and Child 4) using eye gaze to initiate joint attention much more frequently than the other children.

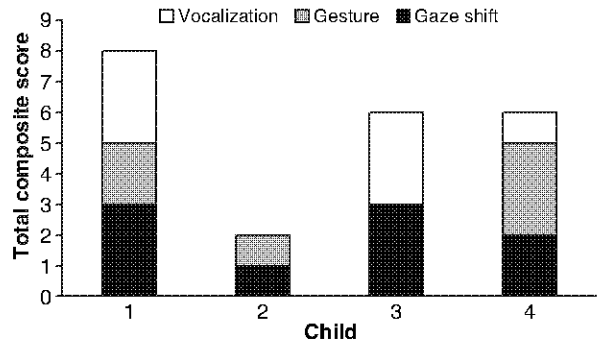

Figure 2 Individual composite scores on joint attention initiation subtests by type of behaviour.

Discussion

This study expands the previous literature on joint attention skills in children with AS, a rare genetic disorder that is associated with severe intellectual disability, motor and speech impairments. Children with AS have more severely impaired expressive communication skills than would be expected on the basis of their level of intellectual functioning alone (Gentile *et al.* 2010). This finding has led to attempts to identify other factors that may be implicated in their expressive communication impairments. Researchers have reported an overlap between AS and features of ASD, particularly in relation to deficits in children's social communication, imitation and joint attention skills. A modified version of a structured assessment protocol that was developed by MacDonald *et al.* (2006) to study children with autism was used in the present study to provide a more in-depth analysis of joint attention response and initiation skills in four children with AS.

All four children in this study were relatively less impaired in responding to the examiner's joint attention bids than in initiating joint attention with the examiner. They all received similar total scores on the joint attention response subtests. Three children showed responsiveness to the examiner's gaze shift alone, whereas the fourth child was responsive to the examiner's gaze shift when it was accompanied by a more overt behaviour in the form of a point. Children were considered to have demonstrated joint attention responding if they followed the direction of the examiner's eye gaze (or eye gaze and point) to look towards a target object. Because it was not possible to evaluate whether children were actually aware of the adult's attention to the object (thus demonstrating true joint attention), a more cautious interpretation of the findings is that they exhibited an early or more basic form of joint attention (Morales *et al.* 1998). These same four children were simultaneously

participating in an intervention study that was geared towards improving their basic learning and functional skills, including their communication and visual attention to people and objects. Thus, it remains to be seen whether children with AS who have not received this type of training will display a similar pattern of results.

Children's scores were more variable for joint attention initiation. Child 1 and Child 4 used gaze shift to initiate joint attention with the examiner much more often than the other children. The former children displayed all three types of joint attention initiation behaviours (eye gaze, gestures, vocalizations), whereas Child 2 and Child 3 showed two of the three behaviours. Child 4 in particular used more advanced forms of gestures such as pointing to herself and shaking her head. For the joint attention initiation test, children did not have to integrate gestures and vocalizations with eye gaze. According to Mundy *et al.* (2009), alternating gaze may be a more important and useful measure of joint attention initiation than other behaviours such as pointing. Adopting this perspective, Child 1 and Child 4 showed the most advanced abilities in joint attention initiation. These same two children were generally functioning at a higher level in most aspects of their development, suggesting a link between mental age and joint attention abilities. An examination of children's individual characteristics may help explain the pattern of results. Child 2 was the youngest child in the study and had the most pronounced deficits in cognitive, adaptive and language functioning. He also received a classification of autism on the ADOS. Child 4, on the other hand, was the oldest child in the study. She displayed the most advanced skills for the most part and did not show any evidence of autistic symptomatology on the ADOS. Child 1 and Child 3 fell between these two extremes in terms of their developmental scores, with Child 1 tending to receive higher scores on scales measuring visual reception, receptive language and socialization. While both children exceeded the clinical cut-off for communication impairment on the ADOS, only Child 3 exceeded the cut-off for reciprocal social interaction. Consistent with the study by Peters *et al.* 2004, children with the maternal deletion had the most pronounced skill deficits and features of autism, whereas the child with a different molecular classification (in this case, mutation in the UBE3A gene) had the most advanced skills and no features of autism. There is emerging evidence that children with AS who are missing a larger segment of 15q11-q13 are more likely to meet criteria for a diagnosis of autism and have lower cognitive and expressive language abilities than children with AS who have a smaller deletion in this region (Sahoo *et al.* 2006). This finding raises the possibility that genes in addition to UBE3A may play a role in children's intellectual, communication and social development and contribute to the expression of autism (Bonati *et al.* 2007).

In summary, children's ability to respond to joint attention bids appears to be less impaired than their joint attention initiation skills. Other researchers have reported a similar pattern of results among children with autism (Mundy *et al.* 2009). MacDonald *et al.* (2006) found that virtually all the typically developing 2-year-olds (as well as 3- and 4-year-olds) in their study were responsive to an examiner's point during a joint attention response test, whereas 2-year-olds with autism demonstrated a lower rate of responding overall. However, children with autism who were older (4 years of age) when they were assessed displayed a similar level of performance to all age groups of typically developing children. In regard to joint attention initiation, typically developing children aged 2–4 appeared to be much more likely to demonstrate a range of joint attention initiation behaviours than similarly aged children with autism. These differences in favour of the typically developing children were most evident for their use of gestures and verbalizations (making comments or asking questions). Although it is not possible to directly compare the children with AS in the present study with the children with autism in the MacDonald *et al.* (2006) study, their overall pattern of results is similar (better joint attention responding than initiation). Some of the limitations of this study are its small sample size, the assessment of children's joint attention at a single point in time and the inclusion of children who were receiving Applied Behaviour Analysis intervention to improve their ability to attend to people and objects as well as their communication and imitation skills. Future studies should include the full range of molecular subtypes in AS and assess children's joint attention in relation to environmental factors such as different communicative partners and settings. Strengths of the study include the use of a structured assessment protocol that contains objective measures of joint attention behaviours as well as the inclusion of standardized measures of children's cognitive, language and adaptive functioning.

This preliminary study raises a number of questions in relation to the development of joint attention skills in children with AS. For instance, what is the relationship between children's joint attention responding and initiation and aspects of their cognitive, social, language and motor development? Is joint attention related to other

factors, such as children's chronological age and molecular subtype, or their imitation skills? Are deficits in joint attention responsive to intervention?

Research into joint attention skills in children with AS may lead to greater insights into the nature of their communication and social deficits, which in turn can guide the development of intervention plans. Increasing children's social and communicative competence should be the ultimate goal of intervention, and the information that is yielded by a behavioural assessment of joint attention such as in the present study can assist with designing intervention approaches to meet children's unique needs.

Correspondence

Any correspondence should be directed to Jane Summers, Ph.D., Developmental Pediatrics and Rehabilitation, McMaster Children's Hospital, Chedoke Site, Empire Building, Hamilton Health Sciences, P.O. Box 2000, Hamilton, Ontario, Canada L8N 3Z5 (e-mail: jsummers@hhsc.ca).

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