Clinical Focus

A Survey of Expressive Communication Skills in Children With Angelman Syndrome

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Angelman syndrome (AS) results from partial deletion of the 15th chromosome (Knoll et al., 1989) and occurs in approximately 1 per 10,000 live births (Petersen, Brondum-Nielsen, Hansen, & Wulff, 1995). Individuals with AS exhibit a pattern of developmental delays that include feeding problems in infancy, delays in motor development, ataxic movements, seizures, severe to profound mental retardation, and a lack of expressive speech (Williams, Zori, et al., 1995). The purposes of this article are to summarize clinical research on AS, to provide a description of the communication skills of individuals with AS, and to identify strategies and resources for communication intervention. It begins by reviewing the existing literature on the clinical characteristics of individuals with AS, with an emphasis on communication skills. The second part of the article presents results obtained from a survey of 20 families of children with AS on the children's expressive communication skills. The implications of the findings of the survey are discussed relative to the existing literature.

Angelman syndrome (AS) results from partial deletion or dysfunction of the 15th chromosome (Knoll et al., 1989). Angelman syndrome (AS) has been identified in over 600 individuals nationally (Angelman Syndrome Foundation, 1993). This figure may be low, however, as recent estimates suggest an incidence rate of 1 per 10,000 live births, and many cases of AS may be undiagnosed (Petersen, Brondum-Nielsen, Hansen, & Wulff, 1995).

Individuals with AS exhibit a pattern of developmental delays. Some of the disabilities associated with AS are feeding problems in infancy, delays in motor development, ataxic movements, seizures, hyperactivity, severe to profound mental retardation, and a lack of expressive speech. Craniofacial features may include an indentation or flattening of the skull in the occipital region, wide mouth, tongue and mandible protrusion, and widely spaced or irregular teeth. Ninety percent of children with AS have seizures by age 3 years (Angelman, 1965; Clayton-Smith, 1992, 1993; Williams, Hendrickson, Whidden, & Bueher, 1993). Because of frequent laughing outbursts, positive facial affect, and a wide, awkward gait with flexion at the elbows, AS was initially referred to as *Happy Puppet* syndrome by Dr. Harry Angelman, for whom the syndrome is named (Angelman, 1965).

Although the syndrome was first described in 1965 (Angelman, 1965), it is only recently that the developmental sequelae of AS have been studied. Of primary interest to speech-language pathologists, educators, and families is that individuals with AS develop little or no usable speech or vocalizations, and the use of nonspeech communication forms (gestures, manual signs, and pictures) is less sophisticated than would be predicted by receptive and cognitive abilities (Clayton-Smith, 1993; Jolleff & Ryan, 1993).

This article summarizes communication research on children with AS, describes the communication skills of children with AS, and provides suggestions for communication intervention. It begins by reviewing the existing literature on the clinical characteristics of individuals with AS with an emphasis on communication skills. The second part of the article presents information obtained through a survey of expressive communication skills in children with AS. The implications of the findings from the survey are discussed relative to the existing literature. Specifically, the children with AS who served as subjects in the present study showed greater variability in expressive communication skills than described in previous studies. The article concludes with strategies for communication intervention for individuals with AS.

Review of the Literature

Diagnosis

In most cases, individuals inherit 23 chromosomes from each parent, resulting in 23 pairs of chromosomes numbered 1–23. Each chromosome contains two long arms (the q arms) and two short arms (p arms). Some syndromes,

such as Down syndrome (Trisomy 21), result from an extra chromosome so that the 21st chromosome has three chromsomes instead of the usual pair. In the case of syndromes such as Fragile X, or 4q- syndrome, part of the arm of a single chromosome may be deleted. Syndromes may also result from an imprinting defect on a single gene, as occurs in sickle cell anemia. In single gene disorders, information that may result in developmental malformations is passed on when the gene replicates.

Angelman syndrome results from complex genetic processes that are only now beginning to be understood. According to recent research, there are four ways in which Angelman Syndrome may be inherited. According to Williams, Zori, et al. (1995), the most common cause of AS is a large deletion in the long arm of the 15th chromosome in the q11–13 region from the allele (member of the chromosome pair) derived from the mother. This accounts for 73% of the cases of AS. Figure 1 is an illustration of the region of the 15th chromosome where the deletion occurs. If the deletion occurs on the chromosome derived from the father, the child will have Prader-Willi syndrome, which has different sequelae from AS (Knoll et al., 1989). In approximately 2% of cases, there is disruption in the imprinting control center (ICR) of the gene, which causes dysfunction in the 15q11–13 region (see Figure 1). Five percent of children with AS have inherited both chromosomes 15 from their father (uniparental paternal disomy). Children with uniparental paternal disomy appear to have less severe manifestations of the syndrome than children with larger deletions (Williams, Zori, et al., 1995). In the remaining 20%, there is no visible evidence of a chromosomal deletion. At the time of this writing, studies had just been published proposing that the gene responsible for AS, UBE3A, has been isolated (Kishino, Lalande, & Wagstaff, 1997; Matsuura et al., 1997). Routine genetic testing may not detect AS, and the most accurate genetic test is the fluorescent in situ hybridization (FISH) test.

No large-scale epidemiological studies have been published. It has been reported that most individuals identified in North America are white; however, cases of AS have been reported across racial and ethnic groups. There are no data as to the ratio of males to females with AS. Recurrence risk is highest in those families in which no apparent chromosomal abnormality is found (Williams, Zori, et al., 1995).

Diagnostic criteria have been established that may be used to confirm the diagnosis clinically in about 80% of all cases (Williams, Angelman, et al., 1995). Although genetic testing is the most accurate means of diagnosing AS, approximately 20% of the cases are currently diagnosed solely on the clinical characteristics of the syndrome when there is no visible deletion. Table 1 details the consistent and associated clinical characteristics that are used as criteria for the diagnosis of AS. Electroencephalography (EEG) is used to support the diagnosis (Van-Lierde, Atza, Giardino, & Viani, 1990; Williams, Angelman, et al., 1995). Williams, Zori, et al. (1995) have proposed that hyperactivity may be a more consistent feature of the syndrome than inappropriate laughing and that hyperactivity has a negative impact on learning and social communication. Associated characteristics of particular clinical significance to speech-language pathologists are craniofacial characteristics (protruding tongue, prognathy, wide mouth with widely spaced teeth), strabismus (Schneider & Maino, 1993), feeding and swallowing difficulties in infancy, mouthing, and drooling.

Much of the developmental delay observed in individuals with AS may be attributed to neurologic abnormalities; however, there have only been a few cases in which the brain of individuals with AS have been available for postmortem study. Some neurologic abnormalities that have been reported include cerebellar hyperplasia (excessive tissue) (Williams & Frias, 1982) and unilateral temporal hypoplasia (lack of tissue) (Van-Lierde et al., 1990). Other observed neurologic differences include decreased myelization and thinning of the corpus collosum (Buntinx et al., 1995; Zori et al., 1992). The majority of individuals with AS have seizures (approximately 90%), and seizures

FIGURE 1. Genetic classes of Angelman syndrome.

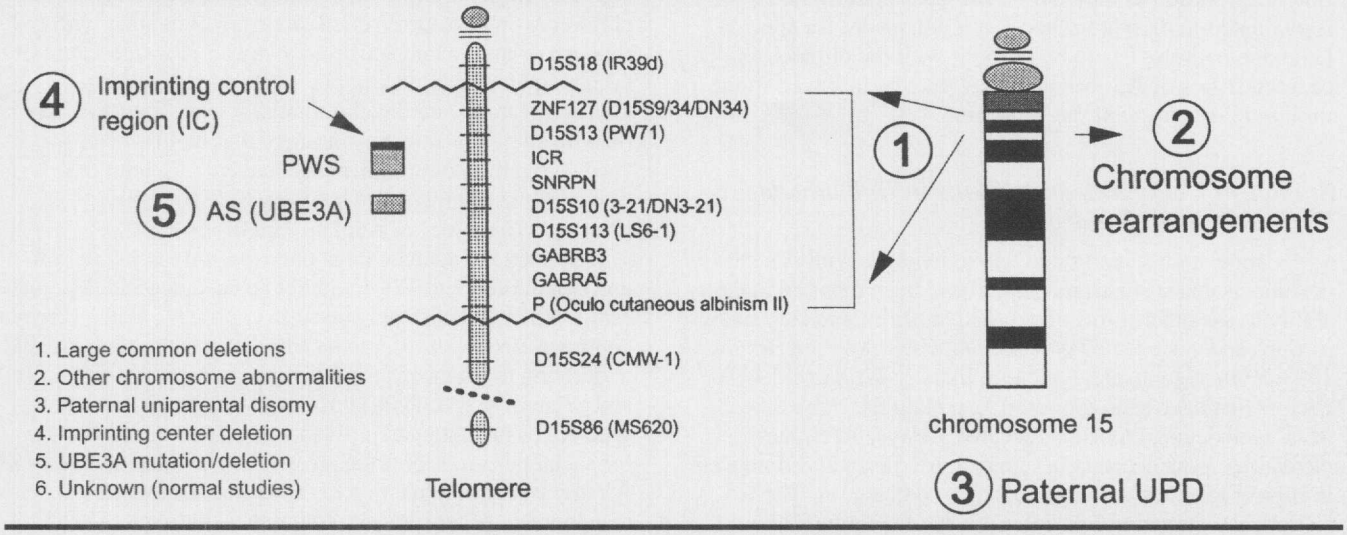

TABLE 1. Clinical consensus criteria for Angelman syndrome.

In 100% of diagnosed cases

1. *Developmental delay:* functionally severe
2. *Speech impairment:* none or minimal use of words; receptive and non-verbal communication skills higher than verbal ones
3. *Movement or balance disorder:* usually ataxia of gait and/or tremulous movement of limbs
4. *Behavioral uniqueness:* any combination of frequent laughing/smiling; apparent happy demeanor; easily excitable personality, often with hand flapping movements; hypermotoric behavior; short attention span

Frequently occurring (more than 80% of diagnosed cases)

1. Delayed, disproportionate growth in head circumference
2. Abnormal EEG, characteristic pattern

Associated (20%–80% of diagnosed cases)

1. Flat occiput, occipital groove
2. Protruding tongue, prognathy, wide mouth
3. Frequent drooling, excessive chewing/mouthing behaviors
4. Hypopigmented skin, hair and eyes (only in deletion cases)
5. Hyperactive lower extremity deep tendon reflexes
6. Uplifted, flexed arm position especially during ambulation
7. Sleep disturbance, attraction to/fascination with water

From: From Williams, Angelman, et al., (1995). Angelman syndrome: Consensus for diagnostic critera. *American Journal of Medical Genetics, 56*, p. 238. (reprinted by permission)

generally occur before age 3. The seizures are generally major motor seizures; however, individuals with AS may experience other types of seizures. Because of seizures, many children receive anticonvulsant medication, and it is unclear how the medications may affect learning in children with AS.

Angelman syndrome is often unrecognized at birth, and diagnosis commonly occurs between the ages of 3 and 7, when behavioral characteristics become most apparent (Williams, Zori, et al., 1995). The most salient characteristics are severe developmental delay; lack of speech; frequent laughter; protruding tongue; a stiff, jerky, and unsteady gait; seizures; hyperactivity; and hypopigmentation of the skin. AS may not be recognized until the child is evaluated in early childhood. It is important for speech-language pathologists to be familiar with the clinical characteristics of AS, because they may be involved in the initial identification of the syndrome.

Studies of Communication Skills in Individuals With Angelman Syndrome

Existing studies report that few individuals with AS develop usable speech, and delays have been reported in the ability to use other forms of expressive communication such as signs and gestures (Clayton-Smith, 1993; Jolleff & Ryan, 1993; Penner, Johnston, Faircloth, Irish, & Williams, 1993). Factors that have been proposed to explain the expressive delay include cognitive and receptive delays, oral motor difficulties, poor social communication skills, and differences in subject selection and data collection procedures. The following section presents descriptions of communication skills in individuals with AS and hypotheses regarding the underlying nature of expressive deficits observed in AS.

Cognitive and Receptive Factors. Much of the delay in expressive communication seen in individuals with AS can be attributed to mental retardation (Williams, Zori, et al., 1995). However, some studies suggest that cognitive delays do not sufficiently account for the expressive speech delays associated with AS.

A study of the communicative abilities of a group of children with AS in the United Kingdom was conducted by Jolleff and Ryan (1993). They assessed 11 children with AS between the ages of 2 years 5 months and 15 years 3 months, including one group of two and one group of three siblings. The investigators used two communication measures to compare speech expression and comprehension and nonspeech expressive and receptive skills. The Receptive-Expressive Emergent Language Scale (REEL; Bzoch & League, 1971) was used to obtain age-equivalent scores for comprehension of spoken language and expressive vocalizations and speech. A modification of the Preverbal Communication Scale (PVCS; Kiernan & Reid, 1987) was used to assess nonverbal receptive and expressive skills. The PCVS assesses the types of gestures used and understood by preverbal individuals.

Using the REEL, the highest age-equivalent obtained by the subjects for receptive language was 22 months (range 9–22 months). The range for expressive communication scores was smaller (6–14 months), and the highest expressive language age was 14 months. Higher expressive scores were associated with the older subjects who were also members of sibling groups. Age-equivalents for expressive communication were less than receptive communication in all but one subject, and the gap between expressive and receptive age-equivalents ranged from 0 months to 11 months.

On the PVCS, the subjects demonstrated the ability to understand simple gestures (open hand to request and looking where someone pointed) but had difficulty imitating or using gestures expressively. The five children in the two sibling groups were able to imitate simple motor acts (clapping, waving), but the other children did not imitate. The authors felt this limited the likelihood of using sign language for expressive communication. Most of the children using gestures relied on physical contact with the referent or interactant (pushing away an unwanted item or taking an adult's hand and placing it on a desired item). Contact gestures are developmentally less sophisticated than distal gestures (e.g., pointing, shaking the head for "no"), which do not require physical contact with the object or the interactant (McClean, McClean, Brady, & Etter, 1991; Werner & Kaplan, 1963).

The authors concluded that the expressive delays exhibited by children with AS could not be accounted for by developmental delays only because expressive skills were not commensurate with receptive skills. They suggested possible underlying mechanisms for the gap between receptive skills and expressive speech: (a) difficulty with the motor act of speech, (b) fundamental problems in using communication for social purposes, or (c) expressive communication skills in AS improving as a function of rather than as a result of development of receptive or cognitive skills.

Penner et al. (1993) evaluated the cognitive, communication, and oral motor skills of 7 adults with AS. All the subjects were institutionalized and had been diagnosed as profoundly retarded. The researchers found that all subjects functioned cognitively within the sensorimotor cognitive period ranging from Stages 2–6. Like Jolleff and Ryan (1993), Penner et al. found that the subjects had difficulty imitating gestures, even when they modeled gestures within the subjects' repertoires. The subjects tended to rely on contact gestures rather than distal gestures. None of the subjects used symbolic communication, such as sign language or speech, though many used gestures such as giving, physical manipulation, or gaze shift from recipient to referent. Penner et al. concluded that there is variation in communication ability among individuals with AS, even among individuals classified as profoundly retarded. Like Jolleff and Ryan (1993), the authors concluded that the lack of cognitive development does not account solely for the lack of speech development.

More research is needed to clearly understand the relationship between cognitive abilities and communication in children with AS. At present, there have been few systematic evaluations of different aspects of cognition, such as attention, memory, and symbolic behavior. Williams, Zori, et al. (1995) have speculated that the attention deficit associated with the disorder, combined with motor deficits, hyperactivity, and poor expressive skills, makes it difficult to get an accurate profile of developmental functioning. However, they speculate that some of the variability seen in the syndrome is related to hyperactivity and that children with better attention skills test in the range of moderate retardation.

Oral Motor Factors. Several authors have suggested that the expressive delay seen in individuals with AS may be attributed partly to structural differences and dysfunction of the oral mechanism (Frias, King, & Williams, 1982; Jolleff & Ryan, 1993; Penner et al., 1993; Williams, Zori, et al., 1995). These authors have reported a number of oral motor problems associated with AS. Infants with AS have difficulty coordinating suck/swallow and have a tongue thrust which makes breast feeding difficult. Many children with AS are bottle-fed, but few require surgical procedures such as a gastrostomy. Early vocal development is characterized by decreased crying, babbling, and cooing. A word-like utterance may develop between 10 and 18 months, but it is not used meaningfully. In early childhood, other oral-motor behaviors develop, such as mouthing and drooling, and these behaviors may last into adulthood.

In their study, Penner et al. (1993) completed oral motor examinations of their 7 adult subjects. Evaluation of the structure and function of the oral mechanism showed that all subjects exhibited mid-facial retrusion with relative prognathism and wide spacing of teeth. Other craniofacial differences were noted, such as macrostomia (excessively wide mouth), and all subjects had an open mouth at rest. Feeding and drooling problems were noted in 6 of the 7 subjects, as were maladaptive oral behaviors such as mouthing of objects and hands, rumination, and pica.

Frias, King, and Williams (1982) reported that the prognathy observed in children with AS may be a secondary effect of excessive chewing and mouthing. The children they have studied have had normal-sized mandibles, but the mandibles had a forward and upward orientation. The mandibular orientation, combined with mid-facial retrusion and a small cranial base, contribute to the prognathy observed in AS.

Social Communication Factors. A finding in most studies of individuals with AS is difficulty in using any communicative form for social communication. It is unclear, however, how much of this delay is the result of experiential factors and how much is characteristic of the syndrome. In the Jolleff and Ryan study, the 5 children in sibling groups were the only subjects who used gestures to communicate, though many of the children had some training using a manual sign system. They reported that their subjects never initiated communication during the clinical trials, and their subjects appeared to use gestures primarily for behavior regulation (requesting), though they did not directly address the issue of communicative functions. Penner et al. (1993) also found the communicative intents used by their subjects were exclusively requests for objects or actions, and none of the subjects were observed to comment or protest; however, some subjects demonstrated the ability to attend jointly to an interactant and object. Only one subject engaged in a reciprocal turn-taking activity. It was hypothesized that the subjects' poor social skills limited their ability to learn language due to decreased opportunities for meaningful communicative interactions.

Subject Selection and Data Collection Factors. Clayton-Smith (1993) described the developmental and clinical characteristics of AS in 82 patients between the ages of 17 months and 26 years in the United Kingdom. The purpose of the study was to identify physical and behavioral characteristics associated with the chromosomal deletion and was not specifically designed to address communication. Information was gained through caregiver interviews, and there were no formal or informal assessment measures of communication. However, Clayton-Smith's study is interesting because her findings suggest that some individuals with AS have more sophisticated expressive skills than those of the subjects described by Jolleff and Ryan (1993) and Penner et al. (1993). Like Jolleff and Ryan, Clayton-Smith found few individuals with AS who used speech to communicate. No subjects spoke more than six recognizable words, and 30% used no speech. However, 20% of the subjects were able to use limited sign language to communicate, some used gestures including pointing, pulling, or pushing, and others used picture boards to communicate. Signs and communication boards are more developmentally advanced forms of communication than gestures, suggesting that some individuals with AS may have a greater ability to use symbolic communication forms than reported elsewhere.

The differences in the findings obtained by experimental studies of Jolleff and Ryan (1993) and Penner et al. (1993) and the survey by Clayton-Smith (1993) may, in part, be explained by differences in the methods of data collection. Jolleff and Ryan elicited gestural behavior in a clinical setting, whereas Clayton-Smith interviewed the families of most of the subjects in their homes. It was

unclear from the Jolleff and Ryan study whether the individuals administering the PVCS and the REEL were familiar to the subjects. If individuals with AS have difficulty using communication for social purposes, it is likely that their performance will be better in a familiar rather than an unfamiliar setting and with familiar rather than unfamiliar interactants.

It is also possible that these initial studies have described a subgroup of individuals with AS. For example, the Penner et al. (1993) subjects were adults that had been diagnosed as profoundly mentally retarded and were institutionalized. Jolleff and Ryan provided only limited case history information and gave no detail as to the type of intervention services their subjects might have received. By contrast, the children with AS with whom the authors of this study had experience were predominantly children who lived at home and had been enrolled in early intervention programs.

Summary. The reports of Jolleff and Ryan (1993), Penner et al. (1993), Williams, Zori, et al. (1995), and Clayton-Smith (1993) are fairly consistent in their descriptions of the communication skills of individuals with AS. There seems to be a consensus among researchers as to an expressive delay that is not solely attributable to cognitive delays and may be related, in part, to oral mechanism dysfunction. Further, Penner et al. (1993) and Williams, Zori, et al. (1995) hypothesize that communication deficits may result from limited opportunities for social interaction.

A Survey of Expressive Communication Skills in Children With Angelman Syndrome

The authors' clinical observations and reports by families of children with AS have indicated that cases reported in the extant literature do not reflect the range of expressive abilities exhibited by children with AS. This is of particular concern because goals and prognostic statements based on expectations derived from the current literature may underestimate the potential for communication development for some individuals with AS.

Methods

In an initial attempt to confirm parental report and the authors' observations, an informal survey of communication skills of children with AS was developed. Because AS is a fairly uncommon disorder, the survey was distributed via the Internet in order to reach individuals across a wide geographical range. The survey was limited to families of individuals with AS who have access to the Internet, and not all those who received the survey via the Internet consented to participate. It should be emphasized that, like other studies of the communication skills of individuals with AS, the results are from an extremely limited, self-selected sample. There are 600 known cases of AS in the United States and an unknown number internationally; at best, the survey represented approximately 2–3% of the known AS cases in the United States. The results of this limited sample should not be considered as representative of individuals with AS but are presented primarily to generate clinical discussion and encourage further clinical research. Substantially larger subject pools would be necessary to determine which factors may be related to the variability in communication skills observed in subjects in the present study.

A questionnaire was developed that elicited information about health, development, communication, and overall satisfaction with professionals (see Appendix A). The researchers subscribed to the Angelman address over the electronic mail system (majordomo@tbag.osc.edu). An introductory letter was sent out to probe for possible interest in participation. The survey was then sent out via e-mail. Respondents were given the option of sending the survey back through electronic mail or printing the survey out, completing it, and returning it through regular postal mail.

Results

Descriptive Information. A total of 20 families of children with AS completed the survey. Respondents included individuals living in Finland (5%, $n = 1$), Canada (15%, $n = 3$), and the United States (80%, $n = 16$). Five respondents were fathers, 12 were mothers; one survey was completed by both parents, one by the child's grandmother, and one by the child's speech-language pathologist with the family's permission. The children with AS ranged from 17 months to 13 years 4 months with a mean age of 6 years 1 month (see Table 2). Seven children were boys, 12 were girls, and for one subject the sex was not reported. Nine children had one sibling, five children had two siblings, one child had three siblings, one child had five siblings, and four children had no siblings. No respondent reported more than one child with Angelman syndrome in a family but one child did have an older sister with spina bifida. All of the children lived with their biological parents at home.

Medical History. Information regarding the children's medical and developmental histories is presented in Table 2. Age at diagnosis ranged from 14 months to 6 years, with one respondent omitting this information ($M = 3$ years 7 months). Seizures of varying types were exhibited by 75% of the children. The majority of children who experienced seizures were taking the medication Depakote. No parents reported hearing impairments in their children, but several reported vision problems, including 25% ($n = 5$) with strabismus, 20% ($n = 4$) wearing glasses, one (5%) wearing glasses and an astigmatism, another child with other visual problems, and one 1 (5%) with strabismus and glasses.

Sixty-five percent ($n = 13$) of the children had achieved independent walking. The age the children walked independently ranged from 14 months to 54 months, with a mean age of 32 months. The 35% ($n = 7$) of individuals who were not walking independently ranged in age from 17 months to 8 years 2 months. All of those individuals who were not walking independently were walking with some form of assistance, with the exception of the 17-month-old, who was belly crawling and able to bear weight when held upright. Ninety percent of the children reportedly sat unsupported.

Feeding problems were common, with 75% ($n = 15$) of the respondents reporting that the child with AS had feeding problems that included difficulty sucking as an

TABLE 2. Developmental information.

Subject	Age	Gender	Diagnosis	Age at Diagnosis	Vision	Age Sat Unsupported (months)	Age Ambulatory (months)
1	1;5	F	D	1;2	S	9	NI
2	2;3	F	N	1;6	G	8	NI
3	3;6	F	D	1;8	S	12	NI
4	3;6	F	D	3;0	S	8	24
5	4;1	M	D	4;0	N	8	48
6	5;2	F	D	2;1	N	20	NI
7	5;3	F	N	5;0	O	9	23
8	5;11	F	N	5;6	N	—	21
9	6;0	F	U	4;0	N	24	36
10	6;0	F	N	5;0	N	18	48
11	6;2	F	D	4;0	N	8	48
12	6;9	M	N	6;0	N	8	17
13	6;11	F	D	1;4	G	25	33
14	7;0	M	D	3;0	S	13	NI
15	7;0	M	D	1;3	S	30	40
16	7;0	M	N	2;6	N	10	14
17	7;3	M	D	4;0	S,G	24	54
18	8;2	F	N	6;0	G	11	NI
19	9;0	M	U	6;0	N	10	30
20	13;4	—	D	—	G	—	24

Note. M = Male; F = Female, — = No response; D = Deletion; U = Uniparental disomy; N = No visible deletion; S = Strabismus; G = Glasses; A = Astigmatism; O = Other; N = No problems; NI = Not independently ambulatory

infant, reflux, and food preferences and aversions. One child had a gastrostomy tube at the time of the study and was unable to tolerate liquids orally.

Communication. Communicative modalities and sociocommunicative skills are summarized in Tables 3, 4, and 5. The use of manual signs and AAC devices varied widely (Table 3). Fifty percent (n = 10) of the sample used some form of sign to communicate. Of those who did use sign, 35% (n = 7) used signs spontaneously for functional communication. The number of signs reportedly used by children with AS ranged from 2 to over 200. Only two individuals used more than 10 signs, with one individual using more than 200 and the other using approximately 40 signs. In general, respondents reported that these signs were approximations and difficult to understand due to the children's motor limitations. Twenty percent (n = 4) of the children who used sign gained the listener's attention before signing. According to the respondents, 55% percent (n = 11) maintained appropriate eye contact during communicative interactions. Three respondents reported that their children were very social and maintained a great deal of eye contact. When communicating with their children, 60% (n = 12) of respondents reported they used speech only and 30% (n = 6) used signs in conjunction with speech. No respondent replied that they used sign only and two respondents did not supply this information.

Augmentative/alternative communication (AAC) systems were being used by the majority of individuals with AS. Pictures alone were being used by 25% (n = 5) individuals. Augmentative devices with voice were used alone by 15% (n = 3) of the children and in conjunction with pictures by 30% (n = 6) of the children. Thirty percent (n = 6) were not using any form of augmentative communication.

Gestures were commonly used by the children (Table 4). Fifty-five percent (n = 11) pointed to indicate wants or interests and 75% (n = 15) reached for desired objects. Fifty percent (n = 10) manipulated others' hands by placing them on objects desired and 75% (n = 15) were able to indicate *yes* and *no* through head nods or shakes. Respondents were asked if children rounded their lips to blow to determine whether the children were using a blowing gesture to depict the action of blowing. However, many respondents interpreted this survey item as a question about oral motor skills. Only 15% (n = 3) of the respondents replied that the children could round their lips to blow, the majority reporting that motor problems made this a difficult activity for the children.

All the children with AS vocalized, and 55% (n = 11) of them used some speech (Table 5). The number of words ranged from one to 15, with an average of five words. It was reported that the words were generally approximations, and some children were inconsistent in their production. The majority of the respondents reported atypical voice quality, and rated voice quality as harsh (10%, n = 2), guttural (35%, n = 7), harsh and guttural (5%, n = 1), harsh, nasal, and guttural (10%, n = 2) or normal sounding (35%, n = 7). One respondent stated "other," describing her daughter's voice quality as "deep." All of the subjects reportedly laughed; however, only 11 subjects (55%) reportedly laughed at inappropriate times.

Other. Respondents unanimously reported that professionals were lacking in specific knowledge about AS,

TABLE 3. Use of non-speech symbolic communication.

Subject	Age	Uses Signs	Signs Spontaneously	Number of Signs	Gains Attention Before Signing	Family Signs to Child	AAC
1	1;5	N	—	—	—	N	N
2	2;3	Y	Y	9	N	Y	P,V
3	3;6	N	—	—	—	Y	N
4	3;6	N	—	—	—	N	N
5	4;1	N	—	—	—	—	P
6	5;2	N	—	—	—	Y	P
7	5;3	Y	Y	40	Y	Y	N
8	5;11	Y	Y	200+	Y	Y	P
9	6;0	Y	Y	7	Y	Y	P,V
10	6;0	N	—	—	—	N	P,V
11	6;2	Y	Y	5	N	N	P
12	6;9	Y	Y	8	N	Y	V
13	6;11	Y	Y	4	Y	Y	P,V
14	7;0	N	—	—	—	Y	V
15	7;0	N	—	—	—	N	N
16	7;0	N	—	—	—	N	N
17	7;3	Y	N	3	N	Y	P
18	8;2	N	—	—	—	—	N
19	9;0	Y	N	7	N	Y	V
20	13;4	Y	N	2	N	Y	P,V

Note. Y = Yes; N = No; — = No response; P = Pictures; V = Voice output system

TABLE 4. Use of gestural communication.

Subject	Age	Uses Gestures	Finger Point	Reach	Physical Contact	Nod/Shakes Head	Round Lips to Blow	Eye Contact Maintained
1	1;5	Y	N	Y	N	N	—	N
2	2;3	Y	Y	N	N	Y	N	Y
3	3;6	Y	Y	Y	N	Y	Y	Y
4	3;6	N	N	N	N	Y	N	Y
5	4;1	Y	Y	Y	N	N	Y	Y
6	5;2	N	N	R	N	Y	N	N
7	5;3	Y	Y	Y	Y	Y	N	Y
8	5;11	Y	Y	Y	N	Y	Y	Y
9	6;0	Y	Y	Y	Y	Y	N	Y
10	6;0	Y	N	Y	N	N	N	N
11	6;2	Y	Y	Y	N	N	Y	Y
12	6;9	Y	N	R	Y	Y	N	Y
13	6;11	Y	Y	R	N	Y	N	Y
14	7;0	Y	Y	R	Y	Y	N	Y
15	7;0	N	W	R	Y	Y	N	—
16	7;0	N	N	N	Y	Y	N	N
17	7;3	Y	N	Y	Y	Y	N	N
18	8;2	Y	N	R	Y	Y	N	Y
19	9;0	N	W	R	Y	Y	N	—
20	13;4	Y	Y	N	Y	Y	N	Y

Note. Y = Yes; N = No; — = No response; W = Whole Hand; R = Reaching but no open/close; H = Harsh; N = Nasal; G = Guttural; S = Sounds normal; O = Other; A = Laughs appropriately; I = Laughs inappropriately

although 60% (n = 12) indicated they were happy with the services they received from speech-language professionals. Some also expressed dissatisfaction that diagnosis was delayed due to lack of knowledge by the treating physician. In addition, 60% (n = 12) of respondents reported that they felt the child's communication abilities were underestimated. Generally, the underestimation was in regard to receptive communication rather than expressive communication. Fifteen percent (n = 3) of the children's communication skills were reportedly consistent across environments, although many families reported their children's communication skills were poorer in unfamiliar settings or with unfamiliar people.

TABLE 5. Speech and vocalizations.

Subject	Age	Vocalizes	Number of Words Used	Voice Quality	Appropriate Laughter
1	1;5	Y	0	S	A
2	2;3	Y	4	H	A,I
3	3;6	Y	0	S	A,I
4	3;6	Y	3	O	A,I
5	4;1	Y	15	H,N,G	A
6	5;2	Y	0	H,G	A,I
7	5;3	Y	4	S	A,I
8	5;11	Y	0	S	A
9	6;0	Y	5	S	A
10	6;0	Y	3	S	A
11	6;2	Y	—	G	A,I
12	6;9	Y	3	G	A
13	6;11	Y	6	H,N,G	A
14	7;0	Y	0	G	A,I
15	7;0	Y	—	S	A,I
16	7;0	Y	0	G	A
17	7;3	Y	2	G	A,I
18	8;2	Y	0	G	A,I
19	9;0	Y	6	G	A
20	13;4	Y	1	H	A,I

Note. Y = Yes; N = No; — = No response; H = Harsh; N = Nasal; G = Gutteral; S = Sounds normal; O = Other; A = Laughs appropriately; I = Laughs inappropriately

The majority of children (65%) engaged in some form of symbolic play (n = 13), though little specific information about symbolic play was given. Ninety-five percent (n = 19) of the children had received early intervention services (developmental services before age 3).

Discussion and Clinical Implications

The results of this very limited sample of children with AS suggest that most of the children in the sample fit the clinical profiles described in previous studies with respect to history of seizures, attainment of developmental milestones, age at identification, oral motor difficulties, and expressive communication skills. However, there was a range of ability in expressive communication skills not reported previously in studies of individuals with AS. Specifically, there appears to be a greater range in the use of symbolic communicative forms and communication skills in some children with AS than previously reported in the literature. These findings will be discussed with respect to previous studies of individuals with AS and clinical implications.

Communicative Forms. The findings of the present study are consistent with the findings of previous studies with respect to the limited use of expressive speech by most individuals with AS. For individuals who speak, speech is often described as "imprecise" and "hard to understand." Previous descriptions of oral motor skills and findings from the present study suggest that oral motor difficulties play a role in the expressive speech delay observed in individuals with AS. Respondents in the present study reported problems with feeding and swallowing in their children with AS, as well as voice quality differences. These characteristics, along with the general motoric problems associated with the syndrome, point to oral motor difficulties as a contributing factor in the speech delay. It is interesting to note that respondents to the present survey reported that some of their children used vocalizations meaningfully. Vocalizations were used alone, for example, to gain a listener's attention before signing or to accompany gestures such as repetition of the syllable /ʌ/ in conjunction with reaching for a desired object. Interestingly, only about half of the respondents reported the inappropriate laughing that was initially used to characterize "Happy Puppet Syndrome" by Angelman in 1965. This is consistent with the description by Williams, Zori, et al. (1995).

Previous studies of individuals with AS have reported use of gestures and very limited use of manual signs. Most of the children in the study relied on gestures and limited sign for communication; however, two subjects acquired more than 40 signs. It is interesting to note that over half of the children with AS engaged in symbolic play. Some details were available about the type of symbolic play shown by children with AS. One child reportedly pretended to serve coffee to her family and pretended to be a cat. Interestingly, this was also the subject who used the greatest number of manual signs. Another child pretended to give a pacifier to a Lambchop puppet. Whereas there appears to be a considerable range in ability to use manual communication across individuals with AS, manual communication, whether gestures or sign, appears be the preferred expressive modality for most individuals with AS.

The potential to use a manual communication system depends on a number of factors, including imitation, the requisite cognitive ability to use a symbolic communication system, adequate gross and fine motor skill, and opportunities for the use of signs in communicative exchanges. It seems that all of these factors may contribute to difficulties in the use of sign language in children with AS. The signs of individuals with AS are often described as imprecise, possibly due to the fine and gross motor limitations associated with the syndrome. In addition, Penner et al. (1993) and Jolleff and Ryan (1993) found that even with the requisite sensorimotor cognitive skills, some individuals with AS had difficulty imitating gestures, suggesting that some individuals with AS may have difficulty using sign language due to poor motor imitation skills. Limited exposure to manual signs cannot be ruled out as a factor in limited use of manual signs by most individuals with AS. Only about half of the respondents reported consistent use of sign language in communicating with their children. The relative contribution of each of these limitations may vary across individuals with AS; however, the fact that a small percentage of children diagnosed with AS may acquire a substantial manual sign vocabulary indicates that the use of manual signs for communication may be an option for many individuals with AS.

Some individuals with AS used AAC systems other than sign. There is little published information about the use of augmentative devices by individuals with AS. Although the Clayton-Smith (1993) study reported that some individuals with AS used augmentative communication, neither the

subjects in the Penner et al. (1993) study nor the Jolleff and Ryan (1993) study reportedly used any form of augmentative device. The authors' survey found that approximately half of the subjects used some type of AAC system. Armstrong (1992) presents a case study illustrating how a young child with AS was trained to use a direct-select voice output device (Wolf). She describes how early introduction of AAC was successful with the client, N.M. Specifically, N.M. became more independent in his adaptive play and less dependent on his family for interpreting his communicative attempts. Further research is needed to determine how individuals with AS may effectively use AAC. However, if the primary problem underlying the lack of speech in AS is a motor problem, then some children with AS may be good candidates for AAC devices.

Receptive Communication. Studies of communication in individuals with AS have focussed on expressive communication skills, and the present study did not directly address receptive skills. Little is known about the range of receptive skills of individuals with AS or whether individuals with AS show preference for certain receptive modalities (sign as opposed to speech).

It is interesting that none of the parents reported hearing problems with their children. One of the authors met with a number of families of individuals with AS, and almost all of the parents reported that their children had experienced chronic otitis media. Further research is necessary to document otitis media in AS as well as the physiological mechanisms that may lead to chronic otitis media. The potential contribution of otitis media to communication difficulties experienced by individuals with AS should not be overlooked.

Communication Skills. Previous studies of individuals with AS have reported poor use of expressive communicative forms for participation in communicative exchanges. The present study found that the social communication skills in some children with AS are better than is reported elsewhere. For example, some of the children gained listener attention before requesting or commenting. However, most of the respondents reported that their children's skills were poorer with unfamiliar interactants and in unfamiliar contexts. This may partially explain the limited use of communication for social purpose reported in previous studies. The ability to participate in communicative exchanges varies among children with AS, and some children do develop the ability to use communicative forms to express a variety of communicative intents and participate in social interactions.

Implications for Intervention for Children With AS

Although the results of the survey described above must be interpreted conservatively, they generally support the previous studies of Clayton-Smith (1993), Jolleff and Ryan (1993), and Penner et al. (1993). However, several differences emerged. The range of communicative abilities found in a small number of children with AS in the present survey exceeded those reported in previous studies. These results suggest that a small group of children with AS may have more potential for symbolic and social communication than previously reported. There may be several reasons for the differences, including sampling considerations, contextual considerations, and experience in early intervention services. Further study and documentation of the range of abilities exhibited by individuals with AS will facilitate diagnosis, assessment, and intervention.

Of primary concern for many families, teachers, and speech-language pathologists is the choice of communicative modality. There are many criteria to consider, including the individual's level of symbolic development, vision and motor skills, availability of communicative partners, and client and family preferences. Families and clinicians have a number of options, and clinicians and families should work together to develop communication systems that are both useful and practical. At present, it appears that the majority of individuals with AS will not develop usable speech or will be unable to rely on speech as their primary communicative mode. The speech-language pathologist may be involved in developing oral motor goals to improve feeding and swallowing and decrease drooling. However, for a very limited number of individuals with AS, the improvement of speech may be an appropriate goal. For individuals at the pre-symbolic level, intervention should focus on the use of gestures and vocalizations, with the ultimate goal being the use of manual signs and/or a symbolic communication system. Some clinicians and families have reported success with graphic systems such as picture exchange systems, as well as devices such as the Alpha Talker. Interdisciplinary teams who choose to use AAC devices with individuals with severe and profound handicaps may avail themselves of the considerable clinical data available.

Individuals with AS exhibit some unique characteristics, such as difficulty in the use of expressive speech, but intervention for individuals with AS is not that different from intervention for individuals with other disabilities. Communication intervention should serve to improve communication development vertically by helping the individual with AS to achieve the highest level of communication development along the continuum from pre-symbolic to symbolic.

In addition, clinicians must attend to what Halliday (1975) described as the horizontal aspects of communication: the use of communicative forms across a range of communicative functions or intents. To learn language, individuals must be able to participate in communicative exchanges with a variety of communicative partners. Participation in communicative exchanges requires a variety of skills, including initiation, responding, and the expression of a variety of communicative intents. For individuals with little or no intentional communication, initial communication goals should focus on finding the individual's preferred communicative mode and developing programs that increase intentional communication (Alvares & Sternberg, 1994). Although early studies of the communication of individuals with AS suggested very limited social-communicative skills, the present study and clinical observation have shown that some individuals with AS demonstrate considerable communicative competence using gestures, vocalizations, signs, AAC systems, and even speech. Regardless of communicative modality, intervention should focus on increasing the number of communicative partners, enhancing the ability to

participate in communicative exchanges of increasing length, and developing modalities with which to express a variety of communicative intents.

Families are frustrated by professionals' lack of knowledge about AS, and professionals may be frustrated by the lack of available information on AS. Professionals and families should set realistic goals for communication development, but goals should not be mistaken for boundaries.

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