

### Indexing Metadata/Description

- › **Title/condition:** Communication Disorders: Angelman Syndrome
- › **Synonyms:** Angelman syndrome: communication disorders; communication impairment: Angelman syndrome; language disorder: Angelman syndrome; language impairment: Angelman syndrome
- › **Anatomical location/body part affected:** Angelman syndrome (AS) affects the brain, specifically the cerebellum and hippocampus
- › **Area(s) of specialty:** Swallowing and Swallowing Disorders, Pediatric Genetic and/or Neurological Disorders, Learning Disabilities, Infant Feeding and Feeding Disorders
- › **Description:** AS is a rare (approximately 1/15,000) neurogenetic disorder characterized by intellectual disability, seizures, jerky/puppet-like movements, inappropriate laughter, specific facial traits, and communication disorders.<sup>(1,2,3)</sup> AS accounts for 6% of all cases of epilepsy and severe intellectual disability in children<sup>(4)</sup>
- › **ICD-9 code:** 758.3 autosomal deletion syndrome
- › **ICD-10 code:** Q93.5 other deletions of part of a chromosome

(ICD codes are provided for the reader's reference, not for billing purposes)

#### › G-Codes

##### • Swallowing G-code set

- G8996, Swallowing functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals
- G8997, Swallowing functional limitation, projected goal status, at initial therapy treatment/outset and at discharge from therapy
- G8998, Swallowing functional limitation, discharge status, at discharge from therapy/end of reporting on limitation

##### • Motor Speech G-code set

- G8999, Motor speech functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals
- G9186, Motor speech functional limitation, projected goal status at initial therapy treatment/outset and at discharge from therapy
- G9158, Motor speech functional limitation, discharge status at discharge from therapy/end of reporting on limitation

##### • Spoken Language Comprehension G-code set

- G9159, Spoken language comprehension functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals
- G9160, Spoken language comprehension functional limitation, projected goal status at initial therapy treatment/outset and at discharge from therapy
- G9161, Spoken language comprehension functional limitation, discharge status at discharge from therapy/end of reporting on limitation

##### • Spoken Language Expressive G-code set

- G9162, Spoken language expression functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals
- G9163, Spoken language expression functional limitation, projected goal status at initial therapy treatment/outset and at discharge from therapy

#### Authors

**Shifra Weltman, MS, CCC-SLP**  
Cinahl Information Systems, Glendale, CA

**Heather Wiemer, MA, CCC-SLP**  
Cinahl Information Systems, Glendale, CA

#### Reviewers

**Amory Cable, PhD, CCC-SLP**  
American Speech-Language-Hearing  
Association (ASHA)

**Tamara Russell**

**Rehabilitation Operations Council**  
Glendale Adventist Medical Center,  
Glendale, CA

#### Editor

**Sharon Richman, MSPT**  
Cinahl Information Systems, Glendale, CA

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–G9164, Spoken language expression functional limitation, discharge status at discharge from therapy/end of reporting on limitation

• **Attention G-code set**

–G9165, Attention functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals

–G9166, Attention functional limitation, projected goal status at initial therapy treatment/outset and at discharge from therapy

–G9167, Attention functional limitation, discharge status at discharge from therapy/end of reporting on limitation

• **Memory G-code set**

–G9168, Memory functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals

–G9169, Memory functional limitation, projected goal status at initial therapy treatment/outset and at discharge from therapy

–G9170, Memory functional limitation, discharge status at discharge from therapy/end of reporting on limitation

• **Voice G-code set**

–G9171, Voice functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals

–G9172, Voice functional limitation, projected goal status at initial therapy treatment/outset and at discharge from therapy

–G9173, Voice functional limitation, discharge status at discharge from therapy/end of reporting on limitation

• **Other Speech Language Pathology G-code set**

–G9174, Other speech language pathology functional limitation, current status at time of initial therapy treatment/episode outset and reporting intervals

–G9175, Other speech language pathology functional limitation, projected goal status at initial therapy treatment/outset and at discharge from therapy

–G9176, Other speech language pathology functional limitation, discharge status at discharge from therapy/end of reporting on limitation

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G-code Modifier	Impairment Limitation Restriction
CH	0 percent impaired, limited or restricted
CI	At least 1 percent but less than 20 percent impaired, limited or restricted
CJ	At least 20 percent but less than 40 percent impaired, limited or restricted
CK	At least 40 percent but less than 60 percent impaired, limited or restricted
CL	At least 60 percent but less than 80 percent impaired, limited or restricted
CM	At least 80 percent but less than 100 percent impaired, limited or restricted
CN	100 percent impaired, limited or restricted
Source: <a href="https://www.cms.gov/">https://www.cms.gov/</a>	

> **Reimbursement:** Reimbursement for therapy will depend on insurance contract coverage;no specific issues or information regarding reimbursement have been identified

> **Presentation/signs and symptoms:** Newborns with AS typically have no symptoms of the disorder, and studies have indicated that prenatal and birth history is typically normal.<sup>(5)</sup> There might be feeding problems in early infancy. AS is usually diagnosed in late infancy, when developmental delay becomes apparent. Seizures typically begin before the age of 3 years<sup>(5)</sup>

• Consistent features – 100% consistent (all affected persons have these features)<sup>(6)</sup>

- Movement or balance disorder, including ataxia and slow, lurching, jerky, or unsteady limb movements<sup>(6,7,30)</sup>
  - Might be expressed as motor problems such as uncoordinated movements, delayed motor development, asymmetric motor function, and/or trunk weakness<sup>(8)</sup>
  - Children with AS have delayed development of motor skills. On average, they sit at about 12 months, crawl at about 18-24 months, and walk at about 4 years<sup>(5)</sup>
- Frequent laughing and smiling behavior is one of the most salient features of the disorder and is increased (in frequency and duration) during social interactions<sup>(7)</sup>
- Stereotypies, including whole body movements or focal movements such as head shaking, finger wiggling, or hand flapping<sup>(6,9)</sup>
- Severe to profound intellectual disability<sup>(10,11)</sup>
- Hypermotor behavior that tends to decrease with age<sup>(9)</sup>
- Significant speech impairment, absent speech<sup>(6)</sup>
- Frequent features – 80% consistent
  - Microcephaly by age 2 years due to delayed head growth<sup>(6)</sup>
  - Seizures usually starting before age 3 years, onset between ages 1 and 5 years<sup>(12)</sup>
    - As many as 90% of individuals with AS have epilepsy<sup>(4)</sup>
- Associated features – 20-80% consistent
  - Dysmorphic facial features<sup>(13)</sup>
    - Prognathia (i.e., jaw protrusion)<sup>(6)</sup>
    - Wide mouth and wide-spaced teeth<sup>(6)</sup>
    - Protruding tongue<sup>(6)</sup>
    - Almond-shaped eyes<sup>(12)</sup>
- Feeding problems
  - Tongue thrusting/suck and swallowing disorder
  - Gastroesophageal reflux disease (GERD) might develop, leading to aspiration risk and poor weight gain<sup>(12)</sup>
  - Frequent drooling
  - Excessive chewing
  - Abnormal food behaviors (e.g., eating nonfood items)
  - Low birth weight, poor weight gain in first year<sup>(5)</sup>
- Constipation<sup>(12,40)</sup>
- Obesity in older children and adults with AS<sup>(12,40)</sup>
- Strabismus (i.e., abnormal alignment of eyes)<sup>(6)</sup>
- Hypopigmentation of the skin, light hair, and eye color<sup>(6)</sup>
- Hyperactive lower extremity deep tendon reflexes<sup>(6)</sup>
- Uplifted, flexed arm position<sup>(6)</sup>
- Wide gait with pronated ankles (foot rolls inward)<sup>(6)</sup>
- Increased sensitivity to heat<sup>(6)</sup>
- Sleep disturbances<sup>(40)</sup>
- Scoliosis
- Behavioral characteristics
  - Individuals with AS exhibit increased behavioral flexibility(i.e., more typical) as compared to those with nonspecific intellectual disability or autism spectrum disorder (ASD)<sup>(14)</sup>
  - Behaviors that have been documented in individuals with AS include aggression, noncompliance, mouthing of objects, tantrums, and stereotypical behavior<sup>(11)</sup>
  - Inattention, difficulty staying focused<sup>(1)</sup>

- Fascination with water, shiny objects, and crinkly items<sup>(6,15)</sup>
- Autistic features such as attention deficit, hyperactivity, and impaired nonverbal communication patterns<sup>(9,12)</sup>
- Impulsivity is less prevalent than in Down syndrome or nonspecific intellectual disability<sup>(14)</sup>
- Many clinical features change as a child with AS grows older<sup>(4)</sup>
  - Hyperactivity lessens/improves<sup>(4)</sup>
  - Impairment in sleep patterns improve<sup>(4)</sup>
  - Seizures might decrease in frequency as the individual ages, with seizure remission often occurring in the late teen and early adulthood years,<sup>(4,40)</sup> however, epilepsy is the most common cause of hospitalization among individuals with AS across the lifespan.<sup>(40)</sup> Some studies have documented recurrence of seizures in the third and fourth decades of life<sup>(40)</sup>
  - EEG abnormalities are less obvious<sup>(4)</sup>
  - Facial features become more distinctive<sup>(4)</sup>
  - Adults are at risk for developing obesity<sup>(4)</sup>
  - Scoliosis and GERD sometimes worsen with age in individuals with AS<sup>(4)</sup>
- Patients with a mutation on chromosome 15 that is inherited only from the paternal side (2-3% of total cases of AS) exhibit some variation in signs and symptoms of the disorder, including.<sup>(12)</sup>
  - Less severe seizures
  - Higher overall functioning
  - More verbal language (over 3 words)
  - Lower incidence of hypopigmentation and microcephaly
  - High body mass index (BMI)
- Mild forms of the disease often go undiagnosed<sup>(5)</sup>
- Speech and language impairments in AS
  - There is a wide range of speech and language abilities in patients with AS. In most cases of AS, there is a severe speech and language impairment with minimal (i.e., up to 5 or 6 words) to no expressive speech<sup>(6)</sup>
  - Language comprehension has been found to be significantly better than language expression and most patients understand basic commands<sup>(1,12)</sup>
  - While many individuals with AS are completely nonverbal, some studies show improved language expression skills with age, with a maximum vocabulary of 2-3 words in most patients<sup>(1)</sup>
  - Patients with milder forms of the disease might have expressive speech but might still struggle with understanding complex directions and abstract thought<sup>(9)</sup>
  - Of all the communicative functions, 'manding' (i.e., requesting and rejecting) is the most developed in individuals with AS
  - Children with AS have most difficulty with imitative behavior.<sup>(16)</sup> Individuals with AS demonstrate less frequent imitative behavior than individuals with severe mental impairment of other etiologies<sup>(16)</sup>
  - Communicative impairment is most severe in individuals with AS who suffer from seizure disorder as well as those with profound intellectual disabilities (as compared to those with more moderate intellectual disabilities)<sup>(17)</sup>
  - Studies have indicated that the highest age-equivalent scores on standardized language tests for children aged 2-15 years is 22 months receptive age equivalent and 14 months expressive age equivalent. Many children tested, however, scored lower on these measures and received as low as 9 month receptive age equivalent and 6 months expressive age equivalent<sup>(18)</sup>
  - The severe language disorder might be due to cognitive impairment in conjunction with poor oral motor skills<sup>(4)</sup>
  - Intelligibility might be reduced due to speech sound errors caused by oral motor weakness and poor coordination<sup>(4)</sup>

## **Causes, Pathogenesis, & Risk Factors**

### › Causes

- AS is a genetic disorder most often caused by defects on chromosome 15. Specifically, AS can be caused by:

- A deletion of the *UBE3A* gene on chromosome 15 or deletion of the *OCA2* gene. AS is usually (70-75% of cases) caused by a chromosome 15 deletion or mutation inherited from the mother<sup>(7,9,11,12,19)</sup>
- In other cases, patients with clinically diagnosed AS are found to have uniparental disomy (UPD), in which the chromosome defect is inherited from the father.<sup>(10)</sup> Paternal uniparental chromosome 15 disomy accounts for only 2-3% of cases<sup>(3)</sup>
- AS can also be due to an imprinting defect on the *UBE3A* gene<sup>(7)</sup>
- In 12-15% of cases, individuals diagnosed with AS do not have a chromosome 15 abnormality and the cause of the disease is unknown<sup>(3,7)</sup>
- The type of genetic mutation has not been found to significantly correlate to severity of symptoms such as epilepsy and seizures<sup>(3,10)</sup>

#### › Pathogenesis

- When the maternal contribution of the *UBE3A* gene on chromosome 15 is deleted, the functions of the hippocampus and cerebellum are affected, as the maternal allele is the one that is usually expressed in those areas. This has been found to cause seizures and cognitive abnormalities in mice with maternally inherited *UBE3A* mutations or deletions<sup>(2)</sup>
- In addition, abnormal or inhibitory phosphorylation (when a phosphate group is added to a protein or other organic molecule) of calcium/calmodulin-dependent kinase type 2 can be the cause of neurological deficits seen in patients with AS<sup>(2)</sup>

#### › Risk factors

- Genetic inheritance
  - Depending on genetic cause of disease, recurrence risk in families with 1 child with AS can vary. If a child has a de novo (i.e., first occurrence) deletion, risk is only 1%, but if AS is due to a chromosomal rearrangement or small inherited interstitial deletion, the risk can be as high as 50%. Fluorescent in situ hybridization (FISH) analysis of the genes associated with AS in the mother of the affected individual can determine risk in future pregnancies.<sup>(17)</sup> If AS is inherited due to (paternal) UPD affecting chromosome 15, risk might be as low as 1/200, but if it is due to paternal 15;15 Robertsonian translocation (a chromosomal rearrangement), risk can be as high as 100%<sup>(17)</sup>
  - Genetic counseling should be provided to the family of AS patients to determine risk of reoccurrence in children from subsequent pregnancies
- Advanced maternal age
  - Increased risk of UPD AS associated with advanced maternal age<sup>(6)</sup>
- Assisted reproductive technologies (ART)
  - Procedures such as in vitro fertilization (IVF) and intracytoplasmic sperm injection (ICSD) have also been associated with AS (due to imprinting errors)<sup>(6)</sup>

## Overall Contraindications/Precautions

- › The speech-language pathologist (SLP) must be aware of the cultural and linguistic backgrounds of the patient and the patient's family, as these can affect assessment and treatment protocol
- › The SLP should involve family/caregivers as well as the patient's teachers and other therapists in the assessment and treatment process in order to obtain more accurate information and the best results
- › The SLP should observe the patient in a variety of settings as well as conduct interviews with family and caregivers to become aware of the patient's level of communicative functioning and to develop appropriate communication and language goals
- › Constant supervision is crucial as patients with AS have a high tolerance for pain, reduced sense of danger, and preoccupation with water and shiny objects, putting them at risk for drowning and other hazards<sup>(13)</sup>
  - The SLP must be aware of patient's pain and tolerance level as well as attention span, anxiety levels, and any other behavioral issues during assessment and treatment
- › The SLP should complete a thorough review of medical history prior to conducting a speech-language evaluation or treatment
- › See specific **Contraindications/precautions to examination** and **Contraindications/precautions** under **Assessment/Plan of Care**

## Examination

### › **Contraindications/precautions to examination**

- History and testing measures might differ according to the specific age, needs, developmental level, and living environment of the patient
- Standardized measures should be interpreted with caution as they have not been standardized on individuals with AS
- Family members/caregivers should be involved in both assessment and treatment of patients with AS as they have a unique perspective and can provide information on the patient that cannot be obtained through formal or informal measures
- Development of an individualized family services plan (IFSP; ages 0-3) or individualized education plan (IEP; school-aged) in collaboration with the parents/caregivers and school personnel is essential to address communication and/or swallowing problems in the home or school environment

### › **History**

#### • **History of present illness**

- When were symptoms of AS first noticed? Which symptoms were noted and by whom? When was AS first diagnosed? Although features become apparent between the ages of 1 and 4 years, the authors of one study conducted in Turkey found that average age of diagnosis is 6.5 years<sup>(1,10)</sup>

#### – **Mechanism of injury or etiology of illness**

- Which physicians follow and care for the patient? Document the nature and severity of chronic issues related to AS (e.g., scoliosis, seizures, GERD)

#### – **Course of treatment**

- **Medical management:** What is the patient's general health status and issues? What is the status of the patient's ear health and hearing? The following is a short summary of the standard medical management often needed for those with AS:

- Antiepileptic therapy to control seizures and epilepsy might be necessary<sup>(13)</sup>
- Physician might monitor patient for constipation, and laxatives might be necessary<sup>(4)</sup>
- Medication or surgery might be recommended to address GERD
- Behavior therapy along with use of melatonin or sedative medications might be effective in treating sleep disturbances<sup>(13)</sup>
- Behavior therapy along with neuroleptic/antipsychotic medications might be used to treat hypermotor behavior<sup>(9)</sup>
- Orthotic bracing or surgery might be needed to correct orthopedic problems such as pronated ankles or tight Achilles tendons. Surgical rod stabilization might help minimize or prevent scoliosis<sup>(4)</sup>
- Adaptive bottles and nipples might be necessary for infants with AS who have feeding problems<sup>(4)</sup>
- Surgery might be necessary to correct strabismus<sup>(4)</sup>
- **Medications for current illness/injury:** Determine what medications physician has prescribed; are they being taken? Common medications include those to control seizures, such as valproate, clonazepam, nitrazepam, and lamotrigine.<sup>(12)</sup> Chronic constipation might be prevented and treated using polyethylene glycol (MiraLAX) in addition to increased fluid intake.<sup>(12)</sup> Consistent use of sunscreen is vital when outdoors due to hypopigmentation and increased risk of sunburn<sup>(12)</sup>
- **Diagnostic tests completed:** Usual tests for this condition are the following:
  - Genetic testing<sup>(12)</sup>
    - 15w11-13 methylation analysis
    - FISH testing
    - Restriction fragment length polymorphism (RFLP) analysis
    - Genetic screen for *UBE3A* gene mutation
  - Electroencephalogram (EEG) is often abnormal in individuals with AS due to seizure activity
  - Physician might monitor for hypothyroidism using blood testing<sup>(16)</sup>

#### – **Home remedies/alternative therapies:** Document any use of home remedies (e.g., ice or heating pack) or alternative therapies (e.g., acupuncture) and whether or not they help

- In a research study conducted in Canada, researchers found no statistically significant differences between a group of children with AS before and after a yearlong behavioral intervention program (applied behavioral analysis [ABA]). However, there were positive trends observed in the treatment group; hence, researchers recommended further research into the efficacy of behavioral intervention programs for patients with AS<sup>(32)</sup>

- Authors of a study that gathered information about 109 individuals with AS (age 16 or older) from the United States, Puerto Rico, and Canada through phone interviews with parents/caregivers found that music was reported to be independently motivating and “very important” by 90% of the study sample<sup>(40)</sup>
- **Previous therapy:** Document whether patient has had speech, occupational, or physical therapy for this or other conditions and what specific treatments were helpful or not helpful
- **Aggravating/easing factors:** Document any factors that aggravate or ease the patient’s symptoms (e.g., do certain textures of food aggravate feeding issues? Do stimulating environments increase hyperactivity?)
- **Body chart:** Use body chart to document location and nature of symptoms
- **Nature of symptoms:** Document nature of symptoms (e.g., constant vs. intermittent, severity). See *Presentation/signs and symptoms*, above
- **Rating of symptoms:** Use a visual analog scale (VAS) or 0-10 scale to assess symptoms at their best, at their worst, and at the moment (specifically address if pain is present now and how much). FLACC scale can be used for nonverbal patients
- **Pattern of symptoms:** Document changes in symptoms throughout the day and night, if any (A.M., mid-day, P.M., night); also document changes in symptoms due to other external variables, document changes with age
- **Sleep disturbance:** Document number of wakings/night
  - Sleep problems are common among individuals with AS; prevalence is 20-80%<sup>(20)</sup>
  - Most common problems are insomnia and difficulty initiating and maintaining sleep<sup>(20)</sup>
- **Other symptoms:** Document other symptoms patient might be experiencing that could exacerbate the condition and/or symptoms that could be indicative of a need to refer to physician (e.g., dizziness, attention issues, digestive issues)
- **Respiratory status:** Document patient’s respiratory status. Does the patient require supplemental oxygen, have a tracheostomy tube, or need mechanical ventilation?
- **Psychosocial status:** Document psychosocial status and/or behavioral problems
  - Collect behavioral data using parent or caregiver questionnaires and/or observation
    - The Child Behavior Checklist (CBCL) can be used to identify behavioral problems in children aged 1;6-17;11 years using a 3-point scale (0 being that a behavior is not exhibited by child, 2 being that a behavior is frequently exhibited by child)<sup>(18)</sup>
  - Document presence and frequency of behaviors such as:
    - laughing and smiling behavior
    - hand flapping
    - hypermotor behavior
    - inattention
    - fascination with water and/or crinkly items
    - tolerance of pain
    - lack of fear
- In a study conducted in the United Kingdom with 19 children with AS (as well as 19 children with Cri du chat syndrome [CdCS; a genetic disorder marked by intellectual disability and developmental delay] and 15 children with Cornelia de Lange syndrome [CdLS; a genetic disorder marked by intellectual disability and distinctive facial features]), researchers found that significantly more participants with either AS (18/19) or CdLS (15/15) scored above the cut-off for autism spectrum disorder (ASD) (as measured by the Social Communication Questionnaire [SCQ], previously known as the Autism Screening Questionnaire) than children with CdCS (12/19).<sup>(37)</sup> Communication skills of the participants with AS that were used to initiate social interaction were much more impaired and used less frequently than in the participants with the CdCS<sup>(37)</sup>
- In a study conducted in the Netherlands, researchers completed a functional analysis and functional communication training with 3 children with AS confirmed by genetic testing in an effort to reduce challenging behaviors. The challenging behaviors present at baseline included head banging, hitting, kicking, hair pulling, biting, and pinching. The children’s classroom teachers performed a functional analysis to determine the communicative intent behind the challenging behaviors. Once it was determined why each child was using each specific challenging behavior, the teachers implemented functional communication training to replace the challenging behaviors with communicative behaviors. Challenging behaviors were reduced in all 3 participants at the end of the study as a result of the functional communication training, supporting the findings of multiple earlier studies with children with AS.<sup>(39)</sup> SLPs should work

- together with classroom teachers and other professionals on the treatment team to use functional communication as a means to reduce challenging behaviors in children with AS
- In a study conducted in the United States, researchers found that parents of children and adolescents with AS who have sleep disturbances have increased levels of stress<sup>(20)</sup>
- If patient or patient's family reports symptoms of depression, anxiety, or other psychosocial or behavioral disturbance, refer to behavioral specialist or mental health professional for additional assessment
- **Hearing:** Document hearing abilities, including hearing loss, history of ear infections, and use of hearing aids or cochlear implants
- **Barriers to learning**
  - **Are there any barriers to learning? Yes\_\_ No\_\_**
  - **If Yes, describe \_\_\_\_\_**
- **Medical history**
  - **Past medical history**
    - **Previous history of same/similar diagnosis:** Ask about speech and language development. Document if any previous speech/language testing has been completed; if it has, review past reports
    - **Comorbid diagnoses:** Ask patient's family/caregiver about other problems, including diabetes, cancer, hearing loss, psychiatric disorders, and orthopedic disorders
    - **Medications previously prescribed:** Obtain a comprehensive list of medications prescribed and/or being taken (including over-the-counter drugs). See *Medications for current illness/injury*, above
- **Social/occupational history**
  - **Patient's goals:** Document what the patient's caregivers hope to accomplish with therapy and in general. It is important for the treatment team to consider the parent's/caregiver's goals when determining the areas of focus for intervention. In a questionnaire study conducted in the Netherlands in which 77 parents of children with AS reported that their priorities for their child's treatment were communication, recreation, self-care (i.e., continence), motor skills, and academic skills<sup>(38)</sup>
  - **Vocation/avocation and associated repetitive behaviors, if any:** Does the patient attend daycare, preschool, or school? If so, what type of support is in place during daycare/school hours? How does the patient communicate in these settings? Does the patient work in an assisted setting? Does the patient interact with peers? What are some of the patient's interests?
  - **Functional limitations/assistance with ADLs/adaptive equipment**
    - Document if and what type of adaptive equipment the patient is using, such as alternative and augmentative communication (AAC) devices, hearing aids, or glasses. Ask patient's caregivers or family members about the effect of the patient's communication deficits on participation in social and occupational activities
    - Individuals with AS develop some self-help skills, but are typically dependent or require assistance for activities of daily living (ADLs), including dressing, feeding, and toileting<sup>(19,33)</sup>
  - **Living environment:** Document information about the patient's living situation including stairs, number of floors in home, and with whom patient lives, (e.g., caregivers, siblings). With whom does the patient communicate? How does the patient communicate at home and/or at school? Who prepares patient's food?
- **Relevant tests and measures: (While tests and measures are listed in alphabetical order, sequencing should be appropriate to patient medical condition, functional status, and setting)**
  - **Arousal, attention, cognition (including memory, problem solving)**
    - Parent/caregiver interview, patient observations, and assessments that rely on visual response only can provide information on cognitive level and adaptive skills (e.g., feeding, dressing, and toileting)
    - Level of cognitive functioning is usually determined following a thorough psychological or neuropsychiatric examination by a psychologist, psychiatrist, or school psychologist
      - IQ testing
      - Mental scale of the Bayley Scales of Infant Development (BSID-II): This scale gathers information on cognitive, language, social, and personal development and describes behavior qualitatively<sup>(21)</sup>
  - **Assistive and adaptive devices:** Determine if patient uses AAC devices. If so, what kind? How does the device impact communication functioning? Common alternative forms of communication are eye gaze, pictures, and other forms of assisted communication; gestures; facial expressions; and sounds or vocalizations with specific meaning
    - In a study involving 11 children with AS in the United Kingdom, researchers found that, with respect to nonverbal communication, most of the children were able to comprehend gestures, pointing, and object manipulation; however, the only effective method of expressive nonverbal communication was object manipulation<sup>(22)</sup>



–In a study conducted in the United States, a researcher collected data about AAC for children with AS from 122 parents.<sup>(34)</sup> The goal of this study was to determine what types of AAC were accepted and rejected by children with AS and why some were successful and others were not. Researchers found that the AAC method used most frequently that also had the highest level of acceptance (23.1%) was electronic devices. Gestures were the second most commonly accepted method of AAC in this group of children with AS (21%); however, gestures were also found to be the type of AAC with the highest rate of rejection (31.3%). The reason cited by parents as being the top reason for either a child accepting advanced electronic AAC devices was “easy/comes naturally/able.” Examples of other reasons frequently given for accepting an advanced electronic AAC device were “understood by others” and “accessible/available.” The most frequently cited reason for rejecting advanced electronic AAC devices was “fails to understand/value device, unmotivated.” Other frequently given reasons for rejecting an advanced electronic AAC device included “child preferred easier methods” and “limited carryover school to home”<sup>(34)</sup>

• **Oral structure and oral motor function:** Perform oral motor screening and/or exam. Assess strength and movement precision of the articulators, as oral motor deficits might affect feeding and swallowing as well as speech production and expressive language output in children with AS<sup>(22,23)</sup>

–Common facial and oral abnormalities in AS include a wide mouth, tongue protrusion, widely spaced or irregularly spaced teeth, and a pointed chin<sup>(22)</sup>

• **Speech and language examination (including reading)**

–**Speech**

- Complete a phonetic inventory and assess articulation accuracy in single words if patient produces words and/or phrases
- An example of a standardized test of articulation is the Goldman-Fristoe Test of Articulation-3 (GFTA-3) which assesses articulation of consonant sounds in individuals 2-21 years of age<sup>(23,24)</sup>
- For detailed information on organic articulation disorders, see *Clinical Review...Speech Sound Disorders: Articulation Disorder, Organic*; CINAHL Topic ID Number: T708924

–**Language**

- Formal/standardized measures: The following are some examples of standardized measures that might be used with children with AS:
  - Inventory of Potential Communicative Acts (IPCA) can be used to give a description of the different forms of communication as well as speech functions that appear in the child’s repertoire.<sup>(24)</sup> This assessment measure identifies 10 communicative acts and functions, and can be used to plan intervention goals focusing on replacing prelinguistic communication skills with more advanced communicative acts that might be in the patient’s repertoire<sup>(24)</sup>
  - The Verbal Behavior Assessment Scale (VerBAS) can be used to measure communicative functions in individuals of all ages and to categorize them as ‘mands’ (requesting and rejecting), ‘tacts’ (labeling and describing), or ‘echoics’ (imitative behavior)<sup>(24)</sup>
    - Individuals with AS have been found to use more ‘mands’ in communication than other functions
  - The Receptive-Expressive Emergent Language Scale can be used to obtain age-equivalent scores for receptive and expressive language abilities in children up to 3 years of age<sup>(23)</sup>
- Informal/observational measures
  - Collect language sample if appropriate or ask parents to provide list of child’s receptive and expressive vocabulary. Most individuals with AS are nonverbal or have a vocabulary of less than 3 words<sup>(24)</sup>
  - Note child’s interaction and play skills. Does he or she engage in dramatic play? Does he or she engage in joint attention? Assess patient’s use of intentional eye gaze (i.e., gaze shifting between an object and an adult can be considered intentional communication or joint attention)
  - Note what methods child uses to gain attention or make requests. Does he or she use gestures? Words? Pictures or objects? Vocalizations? Many studies have found that individuals with AS rarely use manual signs or communicative gestures<sup>(23)</sup>
  - Language comprehension has been found to be significantly better than language expression, and most patients understand basic commands<sup>(4,12)</sup>
  - Children with AS have difficulty with imitative behavior<sup>(25)</sup>
- Systematic preference assessment: in order to identify the items or activities an individual with AS will respond to during therapy (i.e., by making requests for them), it might be necessary to perform a systematic preferences assessment. Systematically present pairs of items (e.g., toy, sippy cup) or activities (e.g., bubbles, peek-a-boo game) in a random

order at least 10 times each to the patient. Document which item the patient reaches for first in each pair to determine which items are “preferred”<sup>(35)</sup>

- For detailed information on developmental language disorders, see the series of Clinical Reviews on this topic
- **Voice:** Assess vocal function, including vocal quality, loudness, pitch, and endurance; vocal quality in individuals with AS is often harsh, guttural, or nasal<sup>(23)</sup>
- If there are specific concerns regarding voice, refer to otolaryngologist for complete workup to rule out laryngeal pathologies prior to completing a full evaluation
- For detailed information on voice disorders, see the series of Clinical Reviews on this topic
- **Fluency:** Rule out or identify atypical speech disfluencies (stuttering)
- For detailed information on disorders of fluency, see the series of Clinical Reviews on this topic
- **Reading:** Complete an assessment of reading skills as indicated, including letter identification, single word reading, and reading comprehension skills, as indicated based on the individual’s age and developmental level
- The vast majority of patients with AS will be unable to read at any level<sup>(18)</sup>
- **Swallow examination:** Assess feeding and swallowing abilities, as many individuals with AS present with feeding problems such as difficulty sucking, reflux (GERD), and food preferences or aversions<sup>(23)</sup>
  - Feeding problems
    - Especially prevalent in newborns due to suck/swallow incoordination; might increase risk of aspiration<sup>(12)</sup>
    - At risk for decreased appetite<sup>(12)</sup>
    - Tongue thrusting<sup>(6)</sup>
    - Frequent drooling<sup>(6)</sup>
    - Excessive chewing or mouthing<sup>(6)</sup>
    - Abnormal food-related behaviors<sup>(6)</sup>
    - Constipation<sup>(6)</sup>
    - Obesity<sup>(6)</sup> in older children
  - For detailed information on assessment and treatment of dysphagia, see the series of Clinical Reviews on this topic
- **Tracheostomy examination:** If present, assess tracheostomy tube and document date of placement, current respiratory status, and use of speaking valve. For detailed information on tracheostomy tubes in children, see *Clinical Review... Dysphagia: Children with Tracheostomy*; CINAHL Topic ID Number: T709082. For detailed information on assessment of a tracheostomy tube and use of a speaking valve, see *Clinical Review... Passy-Muir Tracheostomy & Ventilator Swallowing and Speaking Valve*; CINAHL Topic ID Number: T708919

## Assessment/Plan

### › **Contraindications/precautions**

- **Only those contraindications/precautions applicable to this diagnosis are mentioned below, including with regard to modalities. Rehabilitation professionals should always use their professional judgment**
  - **Due to ataxia and motor coordination problems, patients with this diagnosis are at risk for falls; follow facility protocols for fall prevention and post fall prevention instructions at bedside, if inpatient. Ensure that patient and family/caregivers are aware of the potential for falls and educated about fall prevention strategies** <sup>(8,30)</sup>
  - Patients with AS lack a sense of danger, are often unable to verbalize discomfort and pain, and have a high pain tolerance.<sup>(12)</sup> Therefore, individuals with AS must be carefully supervised throughout life. Risk of various dangers is also high at night as individuals with AS often suffer from insomnia. Caregivers should be aware of fascination with water and high risk for drowning. AS patients should be supervised at all times around water, pools, bathtubs, and even faucets<sup>(12)</sup>
  - Patients with AS have a fascination with shiny, reflective, and plastic objects. Caregivers must supervise patients to make sure patients with AS do not swallow or aspirate small pieces or poisonous particles.<sup>(12)</sup> Caregivers must lock all cabinet doors and watch the patient at all times. Audio and video monitoring is recommended<sup>(12)</sup>
  - Clinicians should follow the guidelines of their clinic/hospital and what is ordered by the patient’s physician. The summary presented below is meant to serve as a guide, not to replace orders from a physician or a clinic’s specific protocols
  - Patients with AS have a high tolerance for pain and increased sensitivity to heat<sup>(6)</sup>
- › **Diagnosis/need for treatment:** Speech therapy is indicated for patients with AS who exhibit a communication disorder

› **Rule out:** AS can be confirmed using genetic laboratory tests in most patients (85%); for the other 15%, a physician must perform a differential diagnosis based on the patient's signs and symptoms.<sup>(4)</sup> A physician must rule out similar conditions:

- ASD

- AS has many features that are similar to those of ASD, including absence of speech, impaired nonverbal communication (such as gestures and facial expression), attention deficit, and delayed motor development.<sup>(9)</sup> However, as opposed to those with ASD, children with AS are interested in social interactions<sup>(9)</sup>

- In a longitudinal follow-up study conducted in the United States of individuals with AS, researchers found that these individuals had more features of ASD and sensory aversions with presence of Class I deletions (approximately 6 million base pairs of DNA) versus Class II deletions (approximately 5 million base pairs of DNA)<sup>(26)</sup>

- Rett syndrome (can be confused for AS, especially in females with AS)<sup>(6,17)</sup>

- Rett syndrome and AS are similar in that children present with developmental delays, severe communication impairments, movement disorders, seizures, autistic behaviors, and physical characteristics such as microcephaly<sup>(27)</sup>

- These two syndromes are caused by mutations to different genes<sup>(27)</sup>

- Prader-Willi syndrome

- Prader-Willi syndrome is the genetic mirror (sometimes called sister syndrome) of AS (both affecting the 15<sup>th</sup> chromosome).<sup>(36)</sup> Cognitive and neurological impairment is more severe in AS

- The risk of epilepsy and febrile seizures is significantly lower for individuals with Prader-Willi syndrome than for individuals with AS<sup>(36)</sup>

- Williams syndrome

- 22q13.3 terminal deletion<sup>(6)</sup>

- Gurrieri syndrome

- Lennox-Gastaut (petit mal variant pattern)<sup>(17)</sup>

- Mowat-Wilson syndrome (has similar facial features, intellectual disability, microcephaly, and seizures as AS)<sup>(6,17)</sup>

- Phelan-McDermid syndrome

- Static encephalopathy with intellectual disability<sup>(6)</sup>

- Cerebral palsy

- Intellectual disability

- Developmental delay

› **Prognosis**

- AS is a lifelong disease for which there is no cure. Patients with AS have a typical life span<sup>(17)</sup>

- Individuals with AS are unlikely to live independently as adults. Various forms of therapy are needed throughout life and can be effective in improving and maintaining motor skills, functional living skills, behavior, and communication

› **Referral to other disciplines:** Refer to dietitian, physical therapist (PT), special education teacher, early intervention specialist, specialty healthcare providers, audiologist, physicians, and behavioral therapist as needed to address associated conditions of AS, including seizures, feeding difficulties, behavior and sleep problems, orthopedic and vision problems, and safety issues.<sup>(12)</sup> Individuals with AS often need ongoing physical therapy to treat orthopedic conditions and ambulation.<sup>(12)</sup>

It is important to maintain contact with primary care providers regarding the patient's needs and progress.<sup>(12)</sup> Parents and caregivers often need to be referred to counseling to help learn to deal with coping with a patient with AS<sup>(9)</sup>

› **Other considerations**

- Early intervention is critical for young patients with AS, especially in the field of speech therapy. Speech therapy should address feeding issues, communication, AAC devices, and verbal/nonverbal forms of communication. Frustration due to inability to communicate needs can lead to behavioral issues, so devising a communication plan is important early on<sup>(12)</sup>

- Concentration and focus tend to improve with age, increasing the ability to learn and acquire new skills even during adulthood. Intervention can be helpful throughout the life of individuals with AS<sup>(12)</sup>

› **Treatment summary**

- Feeding/oral motor

- For patients who are able to follow directions SLPs should develop oral motor goals (to address articulator strength and precision) to help their patients improve feeding and swallowing skills and to decrease drooling<sup>(23)</sup>

- Other modifications might be necessary for safer feeding and swallowing, such as specialized feeding bottle nipples, upright positioning, thickened feedings, and modified food consistencies
- AAC
  - Based on two research studies, individuals with AS might be able to learn to use various forms of AAC such as manual signs, speech-generating devices, and picture-based communication devices<sup>(24)</sup>
    - In the first study, conducted in the United States, researchers surveyed the families of 20 children with AS between the ages of 1.5 and 14 years<sup>(23)</sup>
      - 50% of the children used manual signs, 25% used pictures, 15% were able to use a speech-generating communication device, and 30% did not use any mode of AAC at all
      - Only 55% of the children had expressive speech, with an average vocabulary of 5 words
      - Most of the children used gestures to communicate, with 55% able to point to indicate wants and 75% able to reach for the desired object<sup>(23,24)</sup>
      - The ability to successfully use manual communication systems depends on the subject's cognitive abilities as well as his or her fine and gross motor skills. For example, poor imitation skills or poor fine motor skills might hinder a child's ability to use sign language. These abilities should be taken into consideration when choosing the form of AAC to use
    - In another study conducted in the Netherlands, 79 Dutch children with AS were assessed using the Inventory of Potential Communicative Acts (IPCA)<sup>(24)</sup>
      - The subjects used a variety of communicative forms (gestural communication, eye gaze, verbal communication) and functions, such as social convention (e.g., hello, bye, thanks), attention to self, reject/protest, commenting, choice making, answering, imitation, and requesting an object, action, or information
      - Level of communicative abilities was higher in individuals with AS who lived at home compared to those who lived in a residential facility
      - Approximately 30% of the subjects were able to use some form of AAC, suggesting that the use of AAC is possible with the right supports
  - Multimodal communication: authors of a case study reported on their intervention with a 21-month-old male child with AS in which they used preferred items to teach vocal, gestural, and graphic (picture selection) modes of communication simultaneously to determine the most efficiently learned means of communication for this child. Sessions took place twice weekly for 5 months in his home or his home-based daycare. For the gesture and graphic modes, the child was trained 6 to 12 times per session per mode using the preferred items. The child was not able to imitate vocal sounds (which is typical of AS), so the focus of vocal mode instruction was to increase vocalizations with the goal of vocal imitation. For all modes of communication, the goal was to increase intentional/purposeful communication. Through the first three phases of therapy, it was clear that his most successful mode of communication was graphic and the other two modes were abandoned in order to focus on this mode in the final phases of therapy. By the fifth session, accuracy for the graphic mode with 3 choices of graphic symbols was up to 92%; however, due to a sudden change in health status the study was discontinued at this time after 44 sessions<sup>(35)</sup>
  - In a self-completion survey study conducted with parents of children with AS in the United States, Canada, and the United Kingdom, parents reported frequent use of AAC for communication. Most parents reported that their children relied heavily on the use of unaided, nonsymbolic methods of communication (described below), but there was strong evidence to indicate acceptance of and competence with high-tech AAC devices as well<sup>(31)</sup>
  - For detailed information regarding the use of AAC devices in speech-language therapy, see the series of Clinical Reviews on this topic
- Unaided augmentative communication (such as enhanced natural gestures)<sup>(28)</sup>
  - The authors of a research study conducted in the United States indicated that parents of children with AS find enhanced natural gestures (ENGs) an efficient, relatively easy, and satisfactory method of communication with their children
    - Parents of 10 children with AS (with deletion on the 15th chromosome) were taught to differentiate between natural gestures and ENGs
      - Natural gestures are spontaneous and unprompted behaviors that can be intentional or unintentional (e.g., pushing away a disliked item, laughing in response to an enjoyable activity)
      - ENGs are intentional, recognizable (in context), and do not rely on actual physical contact with the referent (e.g., a swimming stroke motion made by a child who wants to go into the pool)
    - Parents selected five situations in which they would want to teach ENGs, and recorded their child's natural gestures in these situations, especially those gestures that expressed to parents their child's feelings, wants, needs, likes, and dislikes

- Parents were then taught to use four techniques to encourage and teach their child how to use ENGs: environmental sabotage, expectant delay, mand-model, and molding-shaping
  - Environmental sabotage is when a parent moves a desired object away so that a communication opportunity is created
  - When using expectant delay, a parent arranges the environment so that the child needs some assistance, then moves toward the child, faces the child, and quietly gazes at the child with an expectant expression, waiting for the child to exhibit communicative behavior
  - When a caregiver utilizes the mand-model technique, the environment is arranged in a way that encourages the child to communicate, at which point the caregiver ‘mands’—that is, demands—the child to communicate
    - If the child does so, the parent praises the child, but if the child does not exhibit the communicative behavior/gesture, the behavior is modeled
  - The molding-shaping technique is used by placing an object in the child’s hand and then removing the object while helping the child keep his or her hand shape
    - The hand shape then becomes the child’s ENG for a specific object or action
- Caregiver education/improved interpretation of gestures
  - Since most individuals with AS mainly use prelinguistic behaviors such as pointing, giving/handing something, and eye gaze, intervention approaches should train caregivers and communication partners how to recognize, identify, and interpret the individual patient’s prelinguistic communication acts, learning style, and sensory preferences<sup>(24,41)</sup>
  - Intervention strategies that address caregiver identification and response to prelinguistic acts are often used as short-term goals. The long-term strategies typically focus on developing symbolic communication forms
  - Research indicates that communicative functions such as requesting, rejecting, and commenting are the most developed among individuals with AS<sup>(24)</sup>
- General recommendations
  - Clinicians should teach communicative forms in a variety of different settings, and increase the number of the patient’s communicative partners
  - Communication goals should encourage use of intentional communications using whatever mode the patient prefers. Clinicians should promote this type of communication and focus on increasing the length of communicative exchanges between the patient and his or her communicative partners, as well as increasing the types of communicative functions the patient is able to use (e.g., requesting, commenting, refusing)<sup>(23)</sup>
  - In a research study conducted in the United Kingdom analyzing the association between social interactions and smiling and laughing behavior in AS, researchers found that these “happy” behaviors are heightened during social interactions. (Though excessive laughing and smiling are in fact signs of this syndrome, the authors of the study still identify these behaviors as indications of enjoyment and encourage such behaviors at socially appropriate times.) The authors of the study suggest that social interaction itself might be used as positive reinforcement and can be used to promote desired behavior<sup>(7)</sup>

Problem	Goal	Intervention	Expected Progression	Home Program
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<p>Suck/swallow incoordination, risk of aspiration, tongue thrusting</p>	<p>Reduce aspiration risk, safer feeding</p>	<p><b><u>Therapeutic strategies</u></b></p> <p>Feeding modifications<sup>(29)</sup> Specialized feeding with</p> <ul style="list-style-type: none"> <li>•Bottle nipples</li> <li>•Upright positioning</li> <li>•Thickened feedings</li> <li>•Modified food consistencies</li> <li>•Medications (to address GERD)</li> </ul> <p>For detailed information on dysphagia treatment, see the series of Clinical Reviews on this topic</p>	<p>Progress as appropriate per patient’s ability and tolerance</p>	<p>Demonstrate to family/caregivers how to use safe feeding techniques/equipment at home</p>
<p>Lack of communication; difficulty making requests or choices</p>	<p>Use AAC to make requests and improve ability of other communicative functions</p>	<p><b><u>AAC/aided communication</u></b></p> <ul style="list-style-type: none"> <li>•Sign language</li> <li>•Gestures</li> <li>•Picture cards/communication boards</li> </ul> <p>AAC devices such as picture cards or communication boards can be used to signal immediate needs</p> <p><i>See Treatment summary, above</i></p>	<p>Progress as appropriate per patient’s ability and tolerance</p>	<p>Train family/caregivers how to incorporate use of AAC device in the home setting</p>
<p>Lack of communication in patients with AS; difficulty using AAC devices</p>	<p>Individuals with AS will communicate with family, caregivers, and peers using enhanced natural gestures (ENGs)/without the use of AAC devices</p>	<p><b><u>ENGs</u></b> <sup>(28)</sup></p> <p>Parents can be taught to recognize and enhance their child’s natural gestures<sup>(28)</sup></p> <p><i>See Treatment summary, above</i></p>	<p>Progress as appropriate per patient’s ability and tolerance</p>	<p>Family/caregivers are trained to recognize and encourage the use of ENGs in all settings</p>

<p>Lack of communication in patients with AS, behavior problems caused by frustration due to difficulty communicating</p>	<p>Individuals with AS will communicate with family, caregivers, and peers using gestures or verbal language</p>	<p><b>General recommendations</b></p> <ul style="list-style-type: none"> <li>•Train family, caregivers, and communication partners how to recognize, identify, and interpret the patient's prelinguistic communication acts<sup>(24)</sup></li> <li>•Develop symbolic communication forms <ul style="list-style-type: none"> <li>–Target communicative functions such as requesting, rejecting, and commenting, as these functions were found to be the most developed among individuals with AS<sup>(24)</sup></li> </ul> </li> <li>•Teach communication forms in a variety of different settings</li> <li>•Increase the number of communicative partners the child has</li> <li>•Encourage use of intentional communications using whatever mode the patient prefers</li> <li>•Increase the length of communicative exchanges between the patient and his communicative partners, increase the types of communicative functions the patient is able to use (i.e., requesting, commenting, refusing)<sup>(23)</sup></li> </ul> <p>See <i>Treatment summary</i>, above</p>	<p>Progress as appropriate per patient's ability and tolerance</p>	<p>Parent/caregiver education is vital</p>
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## Desired Outcomes/Outcome Measures

- › Improved functional communication
  - Repeat assessment of speech, language, and communication with measures from initial assessment
  - IFSP/IEP review
- › Increased caregiver response to prelinguistic communication acts
- › Improved and safer swallowing
  - Bedside swallow examination
  - Instrumental examination of swallowing
- › Reduced caregiver stress
  - Caregiver feedback

## Maintenance or Prevention

- › AS is a lifelong condition for which there is no cure. However, therapy can reduce and manage symptoms and improve quality of life
- › Parents of a child with AS should be offered genetic counseling before and/or during subsequent pregnancies

## Patient Education

- › See the Angelman Syndrome Foundation Web site, <https://www.angelman.org/>

## Coding Matrix

References are rated using the following codes, listed in order of strength:

<b>M</b> Published meta-analysis	<b>RV</b> Published review of the literature	<b>PP</b> Policies, procedures, protocols
<b>SR</b> Published systematic or integrative literature review	<b>RU</b> Published research utilization report	<b>X</b> Practice exemplars, stories, opinions
<b>RCT</b> Published research (randomized controlled trial)	<b>QI</b> Published quality improvement report	<b>GI</b> General or background information/texts/reports
<b>R</b> Published research (not randomized controlled trial)	<b>L</b> Legislation	<b>U</b> Unpublished research, reviews, poster presentations or other such materials
<b>C</b> Case histories, case studies	<b>PGR</b> Published government report	<b>CP</b> Conference proceedings, abstracts, presentation
<b>G</b> Published guidelines	<b>PFR</b> Published funded report	

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