



## Angelman Syndrome (AS) For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools ([www.negenetics.org](http://www.negenetics.org)).

Angelman Syndrome can be caused by several different genetic mechanisms. There is some correlation between the specific genetic cause and the severity of individual features.

### Physical characteristics and/or symptoms

*Note: not all people with Angelman syndrome will have all of these features.*

#### Common Features (> 80%)

- Normal prenatal and birth history, normal newborn exam
- Developmental delay, beginning between 6 and 12 months
  - Eventually classified as severe impairment in many affected individuals
  - Actual cognitive abilities may be higher than testing indicates because of the attention deficits, hyperactivity, and lack of speech and motor control
- Movement and balance disorders
  - Ataxic gait is common
  - Balance disorder may cause them to lean or lurch forward
  - Tremulous movement of limbs
- Unique behavior
  - Apparent happy demeanor
  - Easily excitable, frequent laughter
  - Often, hand flapping or waving
  - Hypermotoric behavior
- Speech impairment with no words or minimal words
  - Receptive language skills and nonverbal communication skills better than expressive language
- Absolute or relative microcephaly by the age of 2 years
  - Normal at birth but subsequently have slow growth

- Brain is usually structurally normal but may have mild cortical atrophy
- Seizures
  - Usually begin before the age of 3
  - Characteristic EEG pattern, with large amplitude slow-spike waves

## Other Frequent Findings

- Behaviors
  - Drooling, chewing/mouthing behaviors
  - Food related behaviors including eating non-food items
  - Fascination with water and crinkly items
  - Pleasure responses to high pitched tones
- Sensitive to outdoor and indoor temperatures
- Abnormal sleep/wake cycle and less need for sleep
- Weight issues
  - Infants and young children may have feeding problems
  - By late childhood, obesity can develop
- Scoliosis
- Constipation
- Hypopigmentation on skin and eye (ocular albinism)
- Sensitive to sun
- Prognathism

## Recommended Routine Surveillance

- Evaluation of older children for obesity associated with an excessive appetite
- Ophthalmologic and auditory evaluation prior to school
- Surveillance for scoliosis
- Routine dental care

## Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.

If seizures are present, the following seizure action plan may be useful:

<https://www.epilepsy.com/learn/managing-your-epilepsy/seizure-action-plans>

## Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Developmental evaluation
  - Speech therapy with emphasis on nonverbal methods of communication
  - Physical therapy
  - Occupational therapy
- Gastroenterologist:
  - Significant feeding problems may occur
  - Gastroesophageal reflex
- Geneticist / Genetic Counselor
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
  - Clinical trials
- Neurology
  - Individual should have baseline MRI and EEG
  - Monitor seizures
- Ophthalmology surveillance
  - Strabismus
  - Ocular albinism
  - Visual acuity
- Orthopedists
  - Monitor gait impairment
  - Scoliosis
  - Muscular hypotonia

## Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

*My patient \_\_\_\_\_ has been diagnosed with Angelman syndrome. Severe developmental delay, speech impairment, gait ataxia, microcephaly, and behavioral issues characterize Angelman syndrome. Medical complications with Angelman syndrome include management of seizures, feeding difficulties, scoliosis, gastrointestinal reflux, and constipation. Individuals may also lose their speech and ability to walk. Because of these, \_\_\_\_\_ needs the following accommodations.*

## Seven Important Aspects of School Life

[“Angelman Syndrome at a Glance”](#)

will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



## Resources

### Gene Reviews

<https://www.ncbi.nlm.nih.gov/books/NBK1144/>

### Genetic Home Reference

<https://ghr.nlm.nih.gov/condition/angelman-syndrome>