# Angelman Syndrome

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## Common Names

Angelman Syndrome (AS)
Angels
Happy Puppet Syndrome (no longer viewed as an acceptable term)

## Causes/ etiology

For the majority of people with AS, the cause is a deletion in chromosome 15. This is true for about 70% of those diagnosed with AS. Another five to seven percent have a mutation of the chromosomal region in UBE3A. Two to three percent have no deletion or mutation, but the person is still missing the active UBE3A gene. Some have unusual chromosomal rearrangements and for the rest (about 15%), the cause is still unknown.

## Incidence

Angelman syndrome affects about 1 in 12,000 to 20,000 people.

## Characteristics

*Consistent (100%)*

- Developmental delay, functionally severe
- Speech impairment, no or minimal use of words; receptive and non-verbal communication skills higher than verbal ones
- Movement or balance disorder, usually ataxia of gait and/or tremulous movement of limbs
- Behavioral uniqueness: any combination of frequent laughter/smiling; apparent happy demeanor; easily excitable personality, often with hand flapping movements, and frequent smiling, laughing; hypermotoric behavior; short attention span

*Frequent (more than 80%)*

- Delayed, disproportionate growth in head circumference, usually resulting in microcephaly (absolute or relative) by age 2
- Seizures, onset usually < 3 years of age
- Abnormal EEG, characteristic pattern with large amplitude slow-spike waves

*Associated (20 - 80%)*

- Strabismus
- Hypopigmented skin and eyes
- Tongue thrusting; suck/swallowing disorders
- Hyperactive tendon reflexes

- Feeding problems during infancy
- Uplifted, flexed arms during walking
- Prominent mandible
- Increased sensitivity to heat
- Wide mouth, wide-spaced teeth
- Sleep disturbance
- Frequent drooling, protruding tongue
- Attraction to/fascination with water
- Excessive chewing/mouthing behaviors
- Flat back of head
- Smooth palms

**Long term developmental outcomes**

The severity of the symptoms associated with Angelman syndrome varies significantly across the population of those affected. Some speech and a greater degree of self-care are possible among the least profoundly affected. Unfortunately, walking and the use of simple sign language may be beyond the reach of the more profoundly affected. Early and continued participation in physical, occupational (related to the development of fine-motor control skills), and communication (speech) therapies are believed to improve significantly the prognosis (in the areas of cognition and communication) of individuals affected by AS. The life expectancy of people with Angelman syndrome is similar to that of "typical" individuals. The majority of those with AS achieve continence by day and some by night. Many people with AS improve their living skills with support. Dressing skills are variable and usually limited to items of clothing without buttons or zippers. Most adults are able to eat with a knife or spoon and fork and can learn to perform simple household tasks. General health is fairly good and life-span near average. Particular problems which have arisen in adults are a tendency to obesity (more in females), and worsening of scoliosis if it is present. The affectionate nature which is also a positive aspect in the younger children may also persist into adult life where it can pose a problem socially, but this problem is not insurmountable.

**Assessment approaches**

The diagnosis of Angelman syndrome is based on:

- A history of delayed motor milestones and then later a delay in general development, especially of speech
- Unusual movements including fine tremors, jerky limb movements, hand flapping and a wide-based, stiff-legged gait.
- Characteristic facial appearance (but not in all cases).
- A history of epilepsy and an abnormal EEG tracing.
- A happy disposition with frequent laughter

- A deletion or inactivity on chromosome 15 by array comparative genomic hybridization (aCGH) or by BACs-on-Beads technology.

**Interventions and Treatments**

There is no cure or specific therapy for Angelman syndrome. Medical therapy for seizures is usually necessary in order to treat seizures. Physical and occupational therapies, communication therapy, and behavioral therapies are important in allowing individuals with Angelman syndrome to reach their maximum developmental potential.

**Contributions of the School Psychologist**

- The school psychologist (along with the rest of the team) plays an important role in consideration of the child's educational setting that is best suited for them.
- The school psychologist can provide consultation services to teachers and other professionals working with students who have been diagnosed with AS.
- The school psychologist should encourage family involvement.

**Resources for parents, teachers and professionals**

http://www.angelman.org
http://ghr.nlm.nih.gov/condition/angelman-syndrome
http://www.medicinenet.com/angelman_syndrome/page2.htm