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Speech, Language,  
& Hearing Sciences

# Angelman Syndrome

*First described by Dr. Harry Angelman in 1965, Angelman syndrome is a genetic disorder that results in severe developmental delays and neurological impairments.*

## Fast Facts

- Usually caused by **deletion of gene region** on chromosome 15
- Typically **not passed on** from parent to child
- Affects **1 in 15,000** people
- Boys and girls equally affected
- Diagnosed around age 2 to 5
- Occasionally, but infrequently, co-occurs with **Autism Spectrum Disorder**
- Broad range of **different symptoms**

## Common Characteristics

- Developmental **delay\*** (e.g., not walking until several years old)
- Jerky, poorly coordinated **movements\***
- Frequent, unprovoked **laughing and happy demeanor\***
- Severe **speech impairment\***
- Abnormal **patterns in brain activity**
- **Attention deficits** and hyperactivity
- **Learning disabilities**
- Abnormal **sleep patterns**
- **Seizures** (usually starting before 3 years of age)

## Frequent Physical Features

- Wide mouth and widely-spaced teeth
- Excessive drooling and chewing behavior
- Protruding tongue
- Prominent chin
- Flat back of head
- Smaller-than-average head size
- Lack of pigmentation in skin, hair, and eyes
- Abnormal eye alignment
- Curved spine



## Effects on Language and Communication



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- **Speech rarely acquired for use as primary mode of communication**
  - The highest functioning kids with Angelman syndrome will only have a handful of words
- **Ability to understand language is better than ability to produce it**
  - Can follow simple commands
- **Communication primarily done through:**
  - *Nonspecific vocalizations*
  - *Head nods and shakes*
  - *Eye contact*
  - *Alternative communication devices*
    - Example: B with pictures of items (snacks, toys, movies, etc.). Child points to picture to indicate choice.
  - *Gestures*
    - Contact gestures: physical touching object to indicate a want or need
    - Enhanced Natural Gestures: see side box

## What are Enhanced Natural Gestures?

They are gestures that...

- Child already spontaneously produces or is able to produce, given existing motor skills (e.g., lifting and cupping both hands without holding a juice cup)
- Child uses in a communicative way (e.g., to express desire for a juice cup)
- Child does without relying on physical contact with the desired object (e.g., child can replicate gesture of holding the cup without actually holding it)
- Are understood by strangers, given context (i.e., gestures tend to be more literal rather than figurative)

## What to expect from therapists' approach to care:

- Team approach
  - Physical therapist for gross motor movements
  - Occupational therapist for fine motor movements
  - Speech therapist for communication development
  - Including parents in aspects of intervention
  - And more!
- Speech Therapy Goals
  - Maximizing functional communication (e.g., turn-taking, appropriately rejecting undesired object)
  - Gesture training, especially using Enhanced Natural Gestures
  - Picture communication to express choices between objects
- Therapists should maintain high expectations and challenge your child to reach his or her full potential!

FOR CAREGIVER SUPPORT AND MORE INFORMATION, VISIT:

- <http://www.angelman.org/>
- <http://cureangelman.net/>
- <https://rarediseases.org/>
- <http://ghr.nlm.nih.gov/>