Angelman Syndrome is a severe neurodevelopmental disorder caused by deficient expression of the *UBE3A* gene on chromosome 15 (inherited by the mother). The inheritance of Angelman Syndrome is not typically mendelian. Advanced paternal age is a risk factor.

Clinical problems include:

1. Severe developmental delay (and in particular learning difficulties)
2. Seizure disorder
3. Sleep problems
4. Lack of autonomy/participation in society in relation to intellectual disability, communication impairment, behavioral adaptation & motor problems

People with Angelman Syndrome have difficulty achieving coordinated psychomotor skills required for mastering Activity of Daily Living Skills (ADLS) such as self-help & independence in feeding, dressing, and toileting, although there is a wide variation in self-help skills. In addition, many have no sense of danger.

According to Piaget's Developmental theories, people with Angelman Syndrome may not function beyond the **sensorimotor stage** (typical development between birth - 2 years). Cognitive arrest may occur around 24 months, causing **severe to profound intellectual disability**.

General developmental level is arrested at approximately 15 months. All people with Angelman Syndrome require supervision, even as adults.

Families often call their effected loved ones 'angels.'

Some caregivers & researchers suggest the character 'Dopey' in *Snow White and the Seven Dwarfs* is evocative of a person with Angelman Syndrome.

Estimated Prevalence of Angelman Syndrome:

between 1 in 10,000 & 1 in 40,000

Angelman Syndrome is never diagnosed at birth and only rarely in infancy because developmental delays often don't become apparent until around 6-12 months, when parents begin seeking a reason for a child not reaching early motor milestones (e.g. independent sitting) or the appearance of seizures.

90% of cases can be diagnosed through genetic testing.

Common Preferences of People with Angelman Syndrome

- Visual preoccupation with shiny surfaces, edges of objects, spinning objects
- Attraction to water (including sounds of water, hands/feet in water)
- Fascination with crinkly papers/plastics
- Activities & sensory items more preferred than food, drink, tangible items
- Preferred activities may include riding a bus, taking a bath, swimming, interacting w/staff, praise, watching TV, listening to music, looking at pictures, browsing books and magazines
- Preference for foods that don't require much chewing (e.g. bananas)

Profile of Various Therapy Related Skills

- Visual tracking of moving objects often impaired
- Localization of sound sources may be marginally impaired, although it isn't often a functional problem
- Children can develop some skills if they practice, although it rarely generalizes.
- Reciprocal exchange in a rhythmic, dialogue-like fashion is difficult and turn-taking is impaired by impulsivity.
- Orientation to speaker often impaired
- Can often shake or nod their heads yes or no
- People with Angelman Syndrome often invite partners into their activity by taking the person's hand.
- People with Angelman Syndrome can often point, using the whole hand or a single finger
- Perception of touch does not seem to be grossly impaired
- Good skills in spatial navigation and long-term memory

Communication Profile in Angelman Syndrome

Marked absence of expressive speech.

- Most people with Angelman Syndrome have no spoken language.
- Those with a few spoken words do not use speech as a primary means of communication
- Spoken words often poorly articulated.

- Spoken sounds limited to vowels or a few consonants
- Most people with Angelman Syndrome understand many single words, especially those referring to concrete objects
- Comprehension of two word phrases may be limited by attention deficits
- Expressive skills are often multimodal

Communication is most often used to mand for preferred objects or activities, less often to tact.

People with Angelman Syndrome can often learn to choose between communication cards.

Most people with Angelman Syndrome use signs to communicate.

Motor Profile in Angelman Syndrome

Gross Motor developmental level often arrested at **24 months.**
Fine Motor developmental level often arrested at **15 months.**

Cerebellar dysfunction contributes to motor impairment. Alternative locomotion devices like walkers, wheelchairs, and tricycles can enhance mobility.

Oral Motor Profile

- 50% show persistent tongue protrusion and drooling
- Otherwise tongue protrusion & drooling is associated with laughter
- The mouth is almost constantly open
- Oral motor imitation very difficult for people with Angelman Syndrome

Gross Motor Milestones for children with Angelman Syndrome:

- Sitting achieved during second year
- Bottom-shuffling achieved 18-24 months
- Independent walking achieved after age 3-4
- 10% will never walk independently

Motor hyperactivity in children:

- Considered involuntary
- Rhythmic, repetitive, often symmetrical, purposeless movements in fixed &

predictable patterns
- Includes finger wriggling, facial grimaces, bruxism, head shaking/nodding, rocking, jumping, walking back & forth
- Most often hand flapping and waving
- Appear intermittently for periods of up to several minutes
- Often precipitated by fatigue, stress, excitement, or boredom
- Can include manipulations of objects, or chewing/mouthing of objects
- Usually **no treatment is required** because it causes no harm or significant social impairment to the child
- **Common motor compulsions include:** walking through doorways, shutting all the doors in a particular place, spreading feces on walls, eye rubbing, and masturbation; (eye rubbing requires treatment as it can cause keratoconus)

Social Profile in Angelman Syndrome

- Social smiling emerges 4-6 weeks (early/normal age)
- Reflexive laughter/giggling develops soon after
- Early smiling/giggling may foster early social interaction, but may also mask intellectual disability
- **Smiling & laughing prominent in childhood**, a hallmark of the condition
- Laughter/smiling may sometimes indicate anxiety
- People with Angelman Syndrome show marked social disinhibition
- Fear of strangers diminished
- Specific phobias may be present (e.g. crowds, noise)
- Initiate eye contact easily
- Maintains eye contact for short periods due to attention deficits

Aggressive behavior is rare in children and adults. Problematic behavior may serve a communicative function.

People with Angelman Syndrome often have trouble with social control level, social feedback sensitivity, interpretation of peer's behavior or awareness of impact of one's own behavior, but do not have a primary deficit of theory of mind.

Attention, Hyperactivity, & Impulsivity

- Short attention span in children, attention span increases with age
- Alerting attention does not seem impaired
- Orienting attention skills variable
- Executive attention (focused/selective attention) most heavily impaired
- Joint attention and communicative sharing often impaired
- High impulsivity (difficulty waiting their turn or delaying responses)
- Attention and impulsivity most often addressed with behavioral approaches

Children with Angelman Syndrome are **easily excited**, but hyperactivity decreases in teens & adulthood.

Sensory Profile in Angelman Syndrome

Mild or moderate deficits in integration of stationary and kinesthetic information and difficulties in anticipating and adapting to postural balance disturbances.

Lack of or marked decrease in response to commonly painful stimuli.

Enjoyment in active movement can foster sensorimotor integration.

Sleep Problems in Angelman Syndrome

Dyssomnias, parasomnias, and overall severe sleep disturbances of multiple types are especially common in children age 2-6. Sleep problems often resolve in teen years and adulthood. Children with Angelman Syndrome may have a diminished need for sleep.

Problems often include:

- Difficulty settling when tired
- Difficulty initiating and maintaining sleep
- Irregular sleep-wake cycle
- Inappropriate behaviors (screaming/laughing)
- Sleep-related seizure disorder
- Sleep-related movement disorder (sleep onset may be accompanied by sleep starts (stereotyped movements/muscle twitches), night rhythmias)

Behavioral management is recommended for sleep problems. Good sleep hygiene involves:

- Safe, reassuring, comfortable, quiet sleep environment
- Clear and consistent sleep schedule
- Reinforce time cues
- Reduce stimulation
- Tight-fitting sleeping bag
- Objective is often to reach a compromise rather than solve problems completely because sleep problems resolve with age and children's alertness/activity levels are often not impacted by reduced sleep

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Seizures and Other Health Information

Comorbid seizure disorder occurs in 90% of patients with Angelman Syndrome (Dan, 2008, p. 9, 105)

- Seizure onset occurs between 18 months - 4 years (Dan, 2008, p. 16)
- May improve in teen years (Dan, 2008, p. 17, 106) but worsen in adult years
- Seizures often appear in clusters.
- Most common types: myoclonic, generalized tonic-clonic, atypical absence

Hypopigmentation: light skin, reduced retinal pigment compared to the rest of the family

Vision problems are common, but often untreated. Some people with Angelman Syndrome have **severe gastrointestinal problems**, or a tendency to develop **scoliosis** (beginning in adolescence) or **multiple-joint stiffening**, sometimes related to **reluctance to exercise** and **reduced mobility** in teenage and adult years

Disabling **resting tremor** may occur in daylong clusters, resulting in loss of the ability to eat or walk.

Eating Disorders common: increased risk of developing bulimia, pica, & obesity (women especially)

Differential Diagnosis: Autism Spectrum Disorder

Angelman Syndrome may be considered syndromic of autism, autism may be a comorbidity with Angelman Syndrome, or autism may be characteristic of Angelman Syndrome.

Unlike people with autism, people with Angelman Syndrome are often highly sociable from early infancy, and have distinctive facial features and a happy disposition.

Similarities with autism:

Hand-flapping	Absence of speech	Attention deficits
Hyperactivity	Feeding & sleeping problems	Delays in motor development
Impaired use of nonverbal communicative behaviors (facial expression, body posture/gestures to regulate social interaction, decoding of facial expression)		

Effects of Angelman Diagnosis on Parents, Family, & Caregivers

Parents often suffer more from child's sleep disturbances than children, experiencing fatigue, irritability & limitation of activities (Dan, 2008, p. 116).

Parents may have feelings of:

- Uncertainty
- Loss of control
- Low self-esteem (parents who use active problem solving techniques have higher self esteem)
- depression (more of a problem in parents of boys) (Dan, 2008, p. 64)

Resources:

Dan, B. (Ed.). 2008. *Clinics in Developmental Medicine No. 177: Angelman Syndrome*. Mac Keith Press: London.

Dan, B., & Christiaens, F. (1999). Dopey's Seizure. *Seizure, 8*, 238-240. Available at: http://www.angelmanuk.org/downloads/infosheets/Dopeys_seizure_Bernard_Dan_1999.pdf