

## Angelman syndrome characteristics\*

### A. Consistent (100%)

- Developmental delay, functionally severe
- Speech impairment, none 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
- Behavioural uniqueness: any combination of frequent laughter/smiling; apparent happy demeanour; easily excitable personality, often with hand flapping movements; hypermotoric behaviour

### B. Frequent (more than 80%)

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

### C. Associated (20 – 80%)

- Flat occiput
- Occipital groove
- Protruding tongue
- Tongue thrusting; suck/swallowing disorders
- Feeding problems and/or hypotonia during infancy
- Prognathia
- Wide mouth, wide-spaced teeth
- Frequent drooling
- Excessive chewing/mouthing behaviours
- Strabismus
- Hypopigmented skin, light hair and eye colour (compared to family), seen only in deletion cases
- Hyperactive lower limb deep tendon reflexes
- Uplifted, flexed arm position, especially during ambulation
- Wide based gait
- Increased sensitivity to heat
- Sleep disturbance
- Attraction to/fascination with water
- Abnormal food related behaviours
- Obesity (in older child)
- Scoliosis
- Constipation

\* From Williams C.A. et al, Angelman Syndrome 2005: Updated Consensus for Diagnostic Criteria. American Journal of Medical Genetics 1401:413, 2006.



# Angelman Syndrome

## Common characteristics and questions about AS

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**Angelman syndrome (AS) is a genetic disorder first described by an English pediatrician, Dr. Harry Angelman, in 1965. He noted that these children had some traits in common: an unstable jerky gait, unusually happy demeanour, variable developmental delay (usually severe by traditional testing methods), lack of speech/ few words (receptive language skills may be much higher than expressive language skills), small head size, abnormal electroencephalograms, and seizure disorder.**

**Other features often noted are sleep disturbances, flattened back of the head, excessive drooling, chewing, and other oral behaviours, hyperactivity, hypopigmentation when compared to other family members, wide-based gait, and feeding problems in infancy.**

**Angelman syndrome is commonly misdiagnosed. Ten percent of cases have to be determined clinically.**

### **Why are some doctors not familiar with Angelman syndrome?**

Only recently have textbooks in neurology, genetics, and pediatrics begun to reference AS. As familiarity with the disorder increases, many more diagnoses are being made of what was once thought to be an extremely rare condition. It occurs in one person per approximately 25,000 people. Often, AS is undiagnosed or misdiagnosed with cerebral palsy, autism, or other childhood disorders. If you suspect your child has Angelman syndrome, have your physician/health care professional refer you to a Medical Genetics Clinic.

The Canadian Angelman Syndrome Society is dedicated to educating parents and professionals about AS and disseminating information about it. The volunteer board of CASS also organizes and hosts an international conference in Canada every summer in even-numbered years.

### **What is the cause and is testing available?**

Angelman syndrome is caused by a defective gene on the maternally derived chromosome 15q11.2 region. The syndrome occurs with equal frequency in males and females. Approximately 70% of individuals with Angelman syndrome have a chromosome deletion (detected by fluorescence in situ hybridization or F.I.S.H.). Less than 5% have inherited both chromosome 15s from their father (uniparental inheritance) instead of one chromosome 15 from each parent; less than 5% have an imprinting defect in 15q11.2; and 10 to 15% have a small DNA alternation (or mutation) in the Angelman Syndrome Gene or UBE3A. In the remaining AS individuals (approximately 10%), the cause cannot yet be determined by laboratory testing and the diagnosis is based on clinical features. Depending on the cause of AS, families may have from less than 1% (i.e. that of the general population) up to a 50% chance of having another child with AS. Laboratory testing for AS is available at most major medical genetic clinics. Testing often includes a combination of chromosomal and DNA techniques.

### **What kind of research and treatment is being done?**

Angelman syndrome is being researched by numerous professionals in order to describe more accurately its genetics, physical manifestations, and behavioural characteristics. As Angelman children are observed and studied, many educational and behavioural interventions have been shown to be effective in the areas of communication, school related behaviours, compliance, sleep disturbance, and general conduct.

Physical and occupational therapies, speech and language intervention, behaviour modifications, and parent training have been shown to be worthwhile. Studies on therapies for epilepsy and sleep disturbance are ongoing.

Additionally, animal models of AS are being studied to understand the underlying biology and tissue specific expression of critical genes in AS.

