

# Facts About AS

## Developmental and Physical Findings

Consistent (100%)

- Developmental delay, functionally severe
- Movement or balance disorder, usually ataxia of gait and/or tremulous movement of limbs. Movement disorder can be mild. May not appear as frank ataxia but can be forward lurching, unsteadiness, clumsiness, or quick, jerky motions
- Behavioral uniqueness: any combination of frequent laughter/smiling; apparent happy demeanor; easily excitable personality, often with uplifted hand---flapping or waving movements;

hypermotoric behavior

- Speech impairment, none or minimal use of words; receptive and non---verbal communication skills higher than verbal ones

Frequent (more than 80%)

- Delayed, disproportionate growth in head circumference, usually resulting in microcephaly ( $\leq 2$  S.D. of normal OFC) by age 2 years. Microcephaly is more pronounced in those with 15q11.2-q13 deletions
- Seizures, onset usually  $< 3$  yrs. of age. Seizure severity usually decreases with age but the seizure disorder lasts throughout adulthood.
- Abnormal EEG, with a characteristic pattern, as mentioned in the text. The EEG abnormalities can occur in the first 2 years of life and can precede clinical features, and are often not correlated to clinical seizure events.

Associated (20 - 80%)

- Flat occiput
- Occipital groove
- Protruding tongue

Source: 7th edition Facts about Angelman Syndrome by Charles A. Williams, M.D., Sarika U. Peters, Ph.D., Stephen N. Calculator, Ph.D. in 2009

## Facts About AS

- Tongue thrusting; suck/swallowing disorders
- Feeding problems and/or truncal hypotonia during infancy
- Prognathia
- Wide mouth, wide-spaced teeth
- Frequent drooling
- Excessive chewing/mouthing behaviors
- Strabismus
- Hypopigmented skin, light hair and eye color (compared to family), seen only in deletion cases
- Hyperactive lower extremity deep tendon reflexes
- Uplifted, flexed arm position especially during ambulation
- Wide-based gait with pronated or valgus-positioned ankles
- Increased sensitivity to heat
- Abnormal sleep wake cycles and diminished need for sleep
- Attraction to/fascination with water; fascination with crinkly items such as certain papers and plastics
- Abnormal food related behaviors
- Obesity (in the older child)
- Scoliosis
- Constipation