

Angelman Syndrome

This syndrome has been confused with cerebral palsy, autism and other childhood disorders. The clinical characteristics of Angelman Syndrome are:

Consistent (100%)

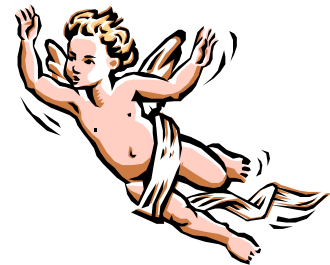
- **Developmental delay, functionally severe**
- **Speech impairment, lack of speech or minimal use of words; receptive & non-verbal communication skills higher than verbal ones**
- **Movement or balance disorder, usually ataxia of gait and/or tremulous movement of limbs**
- **Behavioral uniqueness: any combination of frequent laughter/smiling; apparent happy demeanor; easily excitable personality, often with hand flapping movements; hypermotoric behavior; short attention span**

Frequent (> 80%)

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

Associated (20-80%)

- **Flat occiput (back of the head)**
- **Occipital groove**
- **Protruding tongue**
- **Tongue thrusting; suck/swallowing disorders**
- **Feeding problems during infancy**
- **Prognathia (projecting jaw)**
- **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 limb deep tendon reflexes**
- **Uplifted, flexed arm position especially during ambulation**
- **Increased sensitivity to heat**
- **Sleep disturbance**
- **Attraction to/fascination with water**



It is a genetic disorder caused by abnormal function of the gene UBE3A on chromosome #15. It affects males, females and all racial/ethnic groups equally. There are estimated to be between 1000 and 5000 cases in the U.S. and Canada.

Diagnosis can be made within the first year of birth. It is important to have a physician resource available who is familiar with this disorder. Early diagnosis and treatment are critical. Studies have shown that many educational and behavioral interventions have been effective in areas of communication, schooling, sleep disturbance and general behavior. Physical & Occupational therapies, speech and language interventions, behavior modification and parent training have also been shown to be worthwhile. Alternative/enhanced communication techniques are valuable as children with Angelman Syndrome seem to have greater receptive than expressive language skills.

Resources

Information from : www.angelman.org

Other information can be found by calling, writing or emailing:

1-800-IF-ANGEL

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