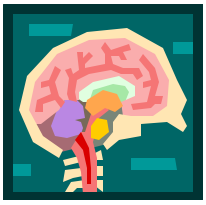

Disability Awareness Begins With You

Angelman Syndrome

What Do We Know?

Angelman Syndrome has confounded and confused the medical community and parents of Angelman individuals for hundreds of years. Initially presumed to be rare, it's now believed thousands of Angelman Syndrome cases have gone undiagnosed or misdiagnosed as cerebral palsy, autism, or other childhood disorders.

Hope Through Research and Recognition

The first glimmer of hope for diagnosis, and ultimately care and treatment of Angelman Syndrome, came as a result of the work and research of Dr. Harry Angelman in 1965. The following are clinical characteristics of Angelman Syndrome:

Consistent (100%):

Developmental delay, functionally severe;

Speech impairment, lack of speech 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;

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 (more than 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.

Today, we know Angelman Syndrome is a genetic disorder caused by abnormal function of the gene UBE3A, located within a small region (q11-q13) on chromosome #15. This region is deleted from the maternally derived chromosome in approximately 80% of individuals with Angelman Syndrome. For the remaining 20%, genetic testing can often identify other abnormalities that disrupt UBE3A function. Some individuals in this latter group, however, still have apparently normal genetic laboratory studies; for these, the diagnosis is based solely upon clinical findings.

Angelman Syndrome 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.

What Can You Do? The Foundation for Knowledge and Connection

Early diagnosis and intervention are beneficial when Angelman Syndrome is suspected. Diagnosis can now be accomplished within the first year after birth. Therefore, a physician familiar with the disorder can be an important resource. The Angelman Syndrome Foundation can provide assistance in connecting families and professionals interested in Angelman Syndrome.

As children with Angelman Syndrome are studied, many educational and behavioral interventions have been shown to be effective in the areas of communication, schooling, sleep disturbances, and general behavior. In addition, physical and occupational therapies, speech and language interventions, behavior modification and parent training have proven worthwhile. A major focus is on alternative/enhanced communication techniques, as children with Angelman Syndrome seem to have much greater receptive language ability than expressive ability.

For more information, call
1-800-IF-ANGEL or write:
Angelman Syndrome Foundation,
414 Plaza Drive, Suite 209
Westmont, IL 60559
Email: info@angelman.org