ANGELMAN SYNDROME FOUNDATION, INC®

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Clinical Features, Diagnosis and Genetics of AS

In this two-part article, you will read about the clinical features and diagnosis or Angelman Syndrome, as well as information on the genetics.

Below is part 1. You can read part 2 here. You'll also find a link to part 2 of this article at the bottom of this page.

Summary of Clinical Features and Diagnosis of Angelman Syndrome

In 1965, Dr. Harry Angelman, an English physician, first described three children with characteristics now known as the Angelman Syndrome (AS). He noted that all had a stiff, jerky gait, absent speech, excessive laughter and seizures. Other cases were eventually published but the condition was considered to be extremely rare and many physicians doubted its existence. The first reports from North America appeared in the early 1980's and within the last seven years many new reports have appeared.

AS has been reported throughout the world among divergent racial groups. In North America, the great majority of known cases seem to be of Caucasian origin. Although the exact incidence of AS is unknown, an estimate of between 1 in 15,000 to 1 in 30,000 seems reasonable.

Developmental and physical features

Angelman syndrome is usually not recognized at birth or in infancy since the developmental problems are nonspecific during this time. Parents may first suspect the diagnosis after reading about AS or meeting a child with the condition. The most common age of diagnosis is between three and seven years when the characteristic behaviors and features become most evident. A summary of the developmental and physical findings has recently been published (21) for the purpose of establishing clinical criteria for the diagnosis and these are listed below. All of the features do not need to be present for the diagnosis to be made, and the diagnosis is often first suspected when the typical behaviors are recognized.

Developmental History and Laboratory Findings

- Normal prenatal and birth history with normal head circumference; absence of major birth defects
- Developmental delay evident by 6 - 12 months of age
- Delayed but forward progression of development (no loss of skills)
- Normal metabolic, hematologic and chemical laboratory profiles
- Structurally normal brain using MRI or CT (may have mild cortical atrophy or dysmyelination)

Consensus Criteria for Clinical Features in Angelman Syndrome

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
- Behavioral uniqueness: any combination of frequent laughter/smiling; apparent happy demeanor; easily excitable personality, often with hand flapping movements; hypermotoric behavior; short attention span

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 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.

You are not alone. There is help. The Angelman Syndrome Foundation is available to provide information, education and support. Our national organization is comprised of regional and local groups that may be close to you. We will strive to help you cope with and understand this special person who has touched your life.

For more information or to obtain free copies of the
Angelman Syndrome tri-fold flyer please contact us at:

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