What is Angelman Syndrome?

Angelman Syndrome (AS) is a genetic disorder first described in 1965 by Dr. Harry Angelman, an English pediatrician. Some common characteristics of Angelman Syndrome are:

- An unstable, jerky gait;
- Unusually happy demeanor;
- 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; and
- Seizure disorders.

Other features often noted are: sleep disturbances, flattened back of the head, excessive drooling, chewing and other oral behaviors, hyperactivity, light skin, eye and hair colors (when compared to other family members), and feeding problems in infancy.

Pacific Northwest

Angelman Syndrome Foundation

1-866-330-9256

representing Alaska, Oregon and Washington

What is PNWASF?

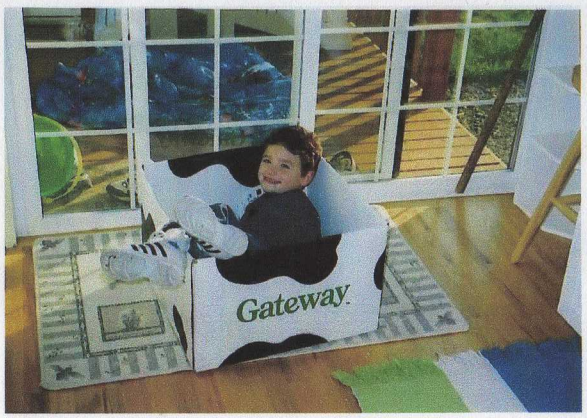
The Pacific Northwest Angelman Syndrome Foundation (PNWASF) was formed in 1994 for the purposes of increasing public awareness of AS. We service as a resource of support for individuals for AS and their families. The organization is comprised of elected officers and directors, and the membership is open to interested individuals, families and professionals. PNWASF is associated with the national Angelman Syndrome Foundations and has contacts with various other national and international AS organizations.

As part of our support role for individuals with AS and their families, PNWASF sponsors camping trips, an adult retreat, regional picnics and holiday functions. The goal of all of these functions is to provide a network of support and information that is so critical to families adjusting to life with an AS child. PNWASF, also, publishes a newsletter and a directory of regional medical professionals familiar with AS, and participates financially and otherwise in the organization of conferences on AS. We have established and are currently maintaining a website. PNWASF is continuing to pursue funding for future research studies related to AS.

What Kind of Research is Being Done?

AS is being researched by numerous professionals in order to more accurately describe its origin, physical signs, symptoms and behavioral characteristics. Genetic studies have identified a tiny deletion in chromosome #15 in approximately 80% of individuals with AS, while in 20% no genetic change has been identified and the diagnosis remains a clinical one. A third very small subgroup has also been identified. These individuals have AS due to the result of receiving two copies of chromosome # 15 from their fathers. This is called "uniparental disomy". Males and females are affected with AS in equal numbers.

As children with AS 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 modifications and parent training have proven worthwhile. A major focus is communication, as children with AS seem to have much greater receptive language ability than expressive ability.



Where Can More Information About AS be Obtained?

Both early diagnosis and intervention can be beneficial when AS is suspected. Diagnosis can be accomplished within the first few weeks of life. Therefore, a physician familiar with the disorder can be an important resource.

PNWASF, with it's network of families in the Pacific Northwest and it's relationships with regional, national, and international AS groups, is available to provide information, education and support to families and professionals interested in AS.

Write or call for more information:

Pacific Northwest Angelman Syndrome Foundation
PNWASF
12932 SE Kent-Kangley Rd., #375
Kent, Washington 98031
1-866-330-9256

Our Website is located at:

http://www.pnwasf.org

Or contact the following local:

David Platts, President at 253-639-1643

Lory Tossey, Vice President at 360-794-4901

Sherri Rein, Treasurer at 253-638-6241