Alexander (Mutation) at ASF AS Clinic, Texas

ANGELMAN SYNDROME

CAUSE | SYMPTOMS

Angelman syndrome is caused by a problem with the UBE3A gene located at the 15th chromosome. It's important to keep in mind that in typical humans, the UBE3A gene from our father is silent and the brain uses the UBE3A gene from our mother during development.

There are 4 ways that Angelman syndrome can occur. These are called genotypes. Each genotype has a different mechanism that results in AS. The 4 types are categorized as: Deletion positive, Mutation, Imprinting Center Defect and Paternal Uniparental Disomy (UPD).

Some symptoms can vary and be more severe than others, but in most children diagnosed with AS, the following are present:

- Developmental delays. These can vary from individual to individual, but common delays are: Infants (0-24 months): inability to support one's head, pull oneself up to stand and delayed motor skills like crawling. Feeding issues due to problems sucking or swallowing. Young children: Delayed ability to walk and an unstable gait or balance issues.
- Seizures. Usually begin to occur between 18 months – 3 years old
- A happy demeanor. Frequent laughing, smiling and easily excitable
- Sleep problems. Abnormal sleep-wake cycles and diminished need for sleep
- Lack of speech. Infants display lack of cooing or babbling; young children usually use nonverbal methods of communication because conversational speech is either absent or limited to very few words.

To request additional information regarding AS you may call: 800-432-6435 or visit Angelman.org.

angelman SYNDROME FOUNDATION | FAMILIES. RESEARCH. CLINICS. COMMUNITY.
WITH YOU FOR THE JOURNEY.