Causes of AS

AS occurs through a genetic mutation, deletion, or missing copies of a gene that produces the UBE3A protein in the brain. In approximately 11% of cases, the cause of AS is unknown.

Incidence

The prevalence of Angelman syndrome is estimated to be approximately 1 in 12,000-20,000 people in the general population. Many cases may go undiagnosed, making it difficult to determine the disorder's prevalence in the general population.

Treatments

Speech Therapy
Behavior Modification
Communication Therapy
Occupational Therapy
Feeding Motility Drugs
Physical Therapy
Special Education
Social Skills Training
Anti-Epileptic Medication

What is Angelman Syndrome?

A complex genetic disorder that primarily affects the nervous system, commonly referred to as AS. Features of this condition include delayed development, intellectual disability, severe speech impairment, and problems with movement and balance.

Features of AS

Angelman syndrome can have a wide variety of symptoms that can vary from person to person. However, children with AS usually experience:

-Delays in reaching milestones
-Severe learning disabilities
-Significant communication deficits.
-A loving, happy, and social demeanor
-Frequent laughter
-Delayed motor development
-Jerky movements
-Stiff-legged walking style
-Balance and coordination difficulties
-Hand flapping
-Hyperactive behavior
-Intellectual disability
-Epilepsy also occurs in more than 80% of cases.

Diagnostic classification

IDEA: Depending on the expression of the disorder, a child with AS may qualify for services under Speech/Language Impairment, Orthopedic Impairment, Intellectual Disability, or Multiple Disabilities

DSM-V: Intellectual Disability

Fun Fact! In general, children with AS have relative strengths in nonverbal reasoning skills and with social interactions in non-verbal contexts.

Assessment approach to AS in schools

Due to intellectual impairment and limited speech abilities associated with AS, it recommended that examiners use assessment measures that have low floors and provide a nonverbal intelligence quotient for evaluations.

Close observation and assessment of the child's sensory-motor functions should be completed because they may be a core deficit requiring intervention through an IEP.

Adaptive skills should also be assessed to determine if skills are intact or need to be included in the IEP.

Tips for Cognitive Assessment:

- Utilize aided language systems/assistive technology to support communication during testing.
- Use alternative assessments that separate motor skill tasks from cognitive tasks to capture what a student with AS understands.
- Utilize portfolio assessment strategies that use multiple forms of data (video footage, sample work, etc.) to measure incremental progress over time.

Long term Developmental outcomes

Possible seizures and aspiration pneumonia.

Accidents due to walking and balance issues and attraction to water that can cause severe injury.

With age, sleep issues and seizures tend to become less severe or infrequent.

Due to mobility issues, obesity and scoliosis can develop in adolescence.

Individuals with AS will require life-long care, but can live long, happy lives.

School Psychologist

We can help ensure students with AS are receiving the most appropriate evidence-based educational interventions to address their multifaceted needs and lead collaborative communication.

Resources

The Angelman Syndrome Foundation's mission is to advance the awareness and treatment of AS for individuals with AS, their families and other concerned parties.
http://www.angelman.org/about-us/

Foundation for Angelman Syndrome Therapeutics (FAST) is an organization of families and professionals dedicated to finding a cure for Angelman Syndrome and related disorders. http://www.CureAngelman.org

Canadian Angelman Syndrome Society (CASS) is dedicated to educating parents and professionals about AS by disseminating information and providing support for parents and caregivers of children with AS.
http://www.angelmancanada.org/