gems genetics education materials for school success

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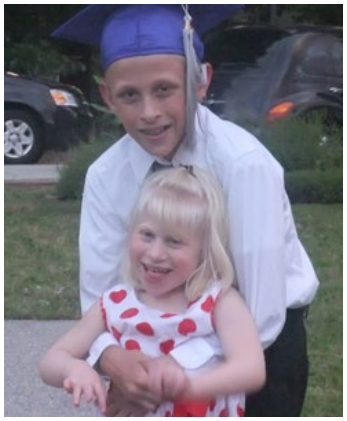
Angelman Syndrome *At a Glance*

Angelman syndrome (AS) is a genetic condition. It is caused by a change on the 15th chromosome. Significant developmental delay or intellectual disability, speech impairment, an abnormal gait, and/or shaking of the limbs are common.
Individuals who have AS also have a unique behavior of *appearing* happy with frequent laughing, smiling, and excitability, often at times not seeming to match the event. Small head size and seizures are also common.



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Angelman syndrome occurs in males and females equally and in all races. About 1 in every 12,000- 20,000 babies is born with AS.

Children who have AS can vary greatly in intelligence, behavior and medical issues.

Things to Think About in the Classroom

1. Medical / Dietary Needs

What you need to know

The list of *possible* medical problems in AS can be quite extensive. However, each individual usually has only *some* of these problems. Also, the severity of any one of these medical problems varies widely. Therefore, it is important to ask the parents about the medical issues for their child.

School age children who have AS should have annual doctor and, often, specialist's visits to monitor medical conditions.

Many children with AS will have seizures. The seizures can be of varying types. The children are very likely to be on anticonvulsant therapy.

Start of puberty, sexual development, and fertility are normal. It is important to provide appropriate sex education

Physical characteristics and/or symptoms:

Not all people with Angelman syndrome have all of these characteristics.

Consistent features: Occur in 100% of children

Developmental delay, functional limitations:

- Actual cognitive abilities may be higher than testing indicates because of the attention deficits, hyperactivity, and lack of speech and motor control
- May differ from others with significant intellectual disability because of their ability to understand some language
- Most need support to live independently as adults

Movement and balance disorders:

- A small number of children are unable to walk (10%)
- Children with AS who learn to walk often start between ages 2.5 and 6 years
- Gait may appear jerky and stiff
- Forearms may be flexed and/or pronated (palms down)
- Tremors occur in limbs
- Movements may be unsteady, clumsy, quick, or jerky. They may tend to lean or lurch forward.
- Increased motor activity is present
- Tongue is often out of mouth; drooling and a wide mouth are present

Unique behavior:

- *Apparent* happy demeanor. This behavior may indicate something other than happiness, including pain.
- Easily excitable, frequent laughter
- Often, hand flapping or waving
- Short attention span
- Some behavior changes may occur due to certain medications

Speech impairment with no words or minimal words:

- Receptive and non verbal communication skills higher than verbal ones
- AAC devices may improve communication

Frequent Findings: Occur in 80% of children

Smaller head:

- Usually have a smaller head

Seizures:

- Seizures: usually start before 3 years of age.
- Seizures become less severe with age but will continue through life
    - Any type of seizure may occur
    - May require multiple medications
    - May be hard to recognize seizures vs. other movements
    - Abnormal EEG

Associated Findings: Occur in 20-80% of children

General:

- Drooling, chewing/mouthing behaviors
- Sensitive to outdoor and indoor temperatures
- If too warm, may be irritable and more active. Skin gets warm.
- Abnormal sleep/wake cycle and less need for sleep
- Fascination with water, crinkly items, papers, and plastic

Food related behaviors:

- May eat nonfood items
- Apparent increased appetite
- High interest in food may lead to obesity

Weight issues:

- Infants and young children may have feeding problems and children with Angelman are often thin and have low subcutaneous fat
    - May be due to poor oral-motor coordination
    - May have gastroesophageal reflux (GERD)
- By late childhood, obesity can develop
    - May occur with age when less mobile and active
    - Ensure activity to help curb obesity

Scoliosis:

- Exercises and activities to help prevent scoliosis may be a part of their physical therapy plan.
- Some children may have alternative therapies, such as chiropractic care, to assist in the treatment of their scoliosis.

Constipation:

- May require a regular laxative

Hypopigmentation on skin and eye (lack of pigment/coloring):

- Sensitive to sun - wear sun protection

What you can do

- Report any change in apparent seizure activity to the parents. Follow school protocols when seizures do occur.
- Seizure activity may be difficult to separate from the child's abnormal movements. If the child is new to your classroom, make sure you speak to the parents about the child's movement disorder and seizure frequency.
- Ensure a yearly check up in the child's Medical Home.
- Ensure up to date immunizations. Most children with AS can receive live virus vaccinations. Record types of vaccinations the child receives.
- Support good hand washing to reduce the spread of viruses.
- Notify parents of changes in energy level.
- Be aware of any changes in behavior or mood. Notify the parents.
- Be aware of any changes in academic performance. Contact parents.
- Be an advocate for the child having communication supports to communicate effectively throughout the day. This may include alternative and augmentative communication systems or devices.
- Dietary: GERD (gastroesophageal reflux) is common. Talk with the parents about particular foods that might be triggers for the reflux and avoid those foods. If the child has more vomiting or reflux than normal, contact the parents so that the cause can be determined

2. Education Supports

What you need to know

It is important to have HIGH EXPECTATIONS for learning for children who have Angelman syndrome. Encourage use of the CORE educational curriculum and modify how it is taught in order to meet the individual needs of the child.

Individualized, flexible, and appropriate educational strategies/supports are keys to success.

Intellectual ability may be underestimated due to lower functional abilities.

- Developmental testing may be difficult because of attention, activity, speech and motor issues.
- Formalized testing has limitations. Make sure testing consists of observations in natural settings.

Children who have AS usually have relative strengths in nonverbal reasoning skills and social interactions.

- Full range of training and enrichment programs should be available.
- Children can do well in regular classrooms when provided with supports needed to be successful.
- The IEP team will help determine what supports and changes are needed.
- Full inclusion with therapies in the regular school setting provides greater learning opportunities.
- It is crucial that children with AS be able to make authentic choices in classroom and life.

Ataxia (difficulty coordinating smooth motor movement)

- Unstable or non-walking children may benefit from physical supports in the classroom.
- They may need extra supports/people to help them in their academic program and inclusion.
- Children who have AS with more motor issues may need extra space and/or minimal obstructions to be safe.
- Physical therapy
    - Adaptive chairs or positioning support may be helpful
- Occupational therapy
    - May help with fine motor and oral motor control
    - Sequencing may be hard due to fine motor challenges.

Attention

- They may pay more attention when they are naturally curious
- High interest is a sign that child is ready to learn sign language and other ways to communicate.

Speech and Language

- Language challenges are significant.

- Use of 1 or 2 words consistently is rare.
    - This may be due to:
        - Motor problems (low tone in oral area)
        - Oral structures (protruding tongue)
        - Intellectual disability
        - Oral apraxia (difficulty with motor planning)
- Most communicate by pointing, using gestures, and using communication boards and AAC devices.
- When children have difficulty communicating, they may resort to pulling hair, hitting, or biting. Make sure they have a communication system that is effective for their needs. Frustration with communication is often the reason for negative behavior. May not need a behavior plan but rather an effective communication system.
- Imitation is difficult. This might not be the best method to teach communication and related skills.
- Maintain high expectations as abilities and methods vary widely.
    - Comprehension is greater than output.

What you can do

Speech and Communication

Teach learning strategies for non-verbal expression.

- Consider new technology, computers, and manual signed systems (ASL, SEE, etc.), depending on fine motor skills. Focus on non-verbal methods of communication such as eye contact, pointing, gesturing, and physical movement.
- When starting to use an AAC device, host a training session for those educators and friends who are most actively involved in the student's life.
- Use augmentative communication aids such as picture cards or communication boards early.
- Communication should work with child's desire to socially interact with others in natural and functional settings such as the home environment, extracurricular settings, and the classroom.
- Make sure children have opportunities for choice and control in their lives and

- Incorporate typically developing peers into their therapy to promote social interaction as well as provide typical models of language.
- If a child uses multiple means of communication, ensure all who work with him have the knowledge of when to use one method vs. another.
- Find an AAC system that allows for maximal social reciprocal communication.
- Anyone interacting with the child should have education and training on how to encourage reciprocal communication with the device.
- When starting to use an AAC device, host a training session for those educators and friends who are most actively involved in the student's life.
- Modeling is important for the use of the AAC device and to encourage its use.
- Continue with strategies that improve oral control to maximize their potential as oral speakers.

Movement

- Physical, speech, and occupational therapy to enable walking, proper positions, hand use, communication needs, etc.
- Bracing and surgery may be needed to align legs.
- Hypermotoric behaviors are often resistant to behavioral therapies.
- Ensure all areas are safe, free of obstacles.

3. Behavioral and Sensory Support

What you need to know

Social

- Young children who have AS learn to respond to personal cues and interactions. They can be very intuitive.
- Interest in people allows children to express a broad range of feelings and form close bonds and real friendships with others.
- They can be part of family and class activities, household chores, and daily living skills.
- They may like recreation, music, and physical activity.

Hyperactivity

- All young children who have AS have a degree of hyperactivity.
    - The constant movement may cause accidental bruises and cuts.
    - Grabbing, pinching, and biting in older children may happen and can be related to this extra movement.
- Behavioral supports may be helpful in limiting the less desirable behaviors that are socially disruptive and self-injurious.
- Some behaviors may be *suggestive* of an Autism Spectrum Disorder. However, social engagement is often quite good, and Autism Spectrum Disorder is not a common diagnosis in children with Angelman syndrome. Some of these behaviors may include:
    - Stereotypic behaviors such as lining up toys, love of spinning
    - Stereotypic motor movements (rocking, hand flapping)
    - Repetitive behaviors and play with unusual objects
    - Sensory interests (lick/mouth/sniff objects)
    - Rituals (hoarding food or objects, food fads)
    - Food related behaviors
        - Eating non-food items
        - Apparent increased appetite
        - Increased interest in food which may lead to obesity

What you can do

Use appropriate teaching strategies.

- Make sure teaching strategies being used are appropriate for children who are already *socially engaged.*

Be proactive with behavioral supports.

- Discuss involvement of behavioral or mental health professionals, or medications with the parents as needed.
- Firm directions, rules, and clear expectations are helpful.

Many children have difficulty regulating emotions and behavior.

- This is especially true when handling unplanned changes.

- Talk through expected changes.
- They usually thrive with consistency and routine. They can be easily upset with disruption.
- Prepare for any change in schedule.
- Provide a safe area to share emotions.
- Teach and model use of words and/or pictures in sharing emotions.
- Teach, emphasize, and reinforce behaviors you want to see.
- Make sure they have an effective communication system.

Provide social cues and coaching.

- Provide information to and discuss differences with the child's peers.
- Help develop confidence and focus on strengths.
- Provide positive reinforcement.
- Teach appropriate social behaviors/skills (e.g., how to ask a friend to play).
- Teach how to recognize facial expressions, body language, and moods in others.
- Teach how to regulate own body – sensory strategies may be helpful.

Behavioral and environmental support for weight management may be needed.

Consider:

- Restricted access to food in all areas
- Locks on the refrigerator
- Constant supervision
- Calorie restriction
- Consistent and scheduled meals and snacks
- Programs that help teach behavioral and weight management strategies.

4. Physical Activity, Trips, and Events

What you need to know

Any change in routine may produce anxiety, fears, and/or worry. Crowds and loud noise may be hard for some children.

If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming (http://nepassage.org/).

What you can do

- Be proactive and discuss any change in schedule or setting with the child ahead of time.
- Use social stories and pictures to help them understand the change.
- Encourage use of their communication system to help them process concerns.

5. School Absences and Fatigue

What you need to know

Sleep issues are common.

- Frequent waking at night
- Difficulty starting or maintaining sleep
- Irregular sleep/wake cycles
- Disruptive night behaviors (i.e., periods of laughter)
- Sleep related seizures

What you can do

- Discuss the child's nighttime sleeping patterns with the parents and provide suggestions on strategies for improvement. Provide consistent routine. Take cues from children who don't have AS.
- Look at temperature in the environment and make it cooler if necessary.
- Schedule daytime naps or afternoon rests if needed.
- Some children respond to a change in scenery (i.e. taking a walk) when tired.
- Lack of restful sleep can contribute to behavior issues and temper tantrums.
- Doctor may discuss:
    - Behavioral treatment programs

- Administration of melatonin
- Use of sedatives

6. Emergency Planning

What you need to know

- Emergency plans will be individually determined, based on behaviors and medical issues.

What you can do

- It is important to mention new signs, symptoms, or pain to the child's parents.

7. Resources

Medline Plus Reference

https://medlineplus.gov/genetics/condition/angelman-syndrome/
Consumer-friendly information about human genetics from the U.S. National Library of Medicine

Angelman Syndrome Foundation

http://www.angelman.org

Our mission is to advance the awareness and treatment of Angelman syndrome through education and information, research, and support for individuals with Angelman syndrome, their families and other concerned parties. We exist to give all of them a reason to smile, with the ultimate goal of finding a cure.

8. Meet a Child with Angelman Syndrome

Meet Ally!

GEMSS would like to thank Ally and her mother for their generosity in sharing this story with us. You have made the site come to life with the addition of your thoughts and feelings. Thank you so much!

Ally has led a busy and fun life in her six years! As a kindergartener in New England (USA), she loves playing with her bother and friends, pretending with her dolls, and watching her favorite princess Belle from Beauty and the Beast.

Ally loves her school and was invited to her first birthday party recently. Her friendships extend beyond the school and it is not unusual for Ally to be approached by friends, whether she is on a playground or picking apples out in the country. She really connects with them and they know her excitement to see them is genuine!

Ally is fully included in her school with therapies woven into the natural routines of the classroom. She uses a communication device which has been a great tool. Her team is continually exploring other communication devices that might broaden her expressive abilities to become more spontaneous and more reciprocal in nature. Ally has thrived in the school with the support and dedication of her team.

Ally's big brother Josh helps program Ally's communication device, lends a hand when she is walking, and plays games with her. But no free ride for Ally – Josh makes sure she plays by the rules of the games!

Ally was diagnosed with Angelman syndrome at 11 months of age after having some feeding difficulties as an infant and, later, with reaching some of her developmental milestones. After a short stay in the hospital, they learned of her diagnosis. She is deletion positive.

Ally's mother advises other parents and teachers to remember that "Kids are kids first, they are not a diagnosis. They have high receptive language skills and understand everything you tell them!" She urges full inclusion and therapies in natural settings. She feels children who have Angelman will thrive by being with other children and learning from real social interactions. She also urges parents and teachers to "go with the strengths of the child – start there in play and use their strengths."