gemss

Genetic Education Materials for School Success

Aicardi Syndrome

Table of Contents

Acknowledgment:

Genetic Education Materials for School Success (GEMSS) was created and maintained by an interdisciplinary team of genetic counselors, physicians, families, and students working with New England Regional Genetics Network (NERGN) and the Institute on Disability at the University of NH. Thank you to these amazing partners. This work was supported by HRSA grant UH7MC30778.

Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

Aicardi Syndrome

At a Glance

Aicardi syndrome is a rare genetic condition found almost exclusively in girls. This condition can vary widely in how it presents. Some girls may have no obvious differences and others may have more significant challenges. Girls with this condition typically have changes in their brain (absence of the *corpus callosum*, which connects the left and right sides of the brain) that can affect speech or movement. They may also have problems with their eyes and often have seizures. Aicardi syndrome is sporadic – it occurs in individuals without a family history of the disorder and can't be predicted.

Meet Joy on page 11.

Common Features of Aicardi Syndrome:

- Seizures – usually difficult to control
- Vision problems
- Moderate-to-severe developmental delay
  - Speech difficulty is common
  -
- Characteristic facial features
  - Short area between upper lip and nose
  - Flat, upturned nose
  - Large ears
  - Sometimes unusual appearance of the colored part of the eye
- Small hands
- Curved spine
- Gastrointestinal problems
  - Constipation, diarrhea, gastric reflux, difficulty feeding

In Depth

Medical and Dietary Considerations

What you need to know

The specific genetic cause of this disorder is not yet known but scientists suspect the X chromosome is involved. The condition occurs almost always in females. In Aicardi Syndrome, the corpus callosum does not develop normally which limits communication across the brain's hemispheres. There may be a variety of other structural and functional changes, but the presence and severity will vary with each child. Medical issues might include some or all of the following:

Seizures

- May have startle seizures in response to sudden sounds
- Multiple medications may be necessary
- Certain diets may help control seizures
- May have a Vagus Nerve Stimulator to help reduce seizure activity
- Anecdotal reports of reduced seizure activity with medical marijuana

Respiratory problems

Children may have congestion which increases risk of infection. Could progress to pneumonia but pneumonia may be due to aspiration

Orthopedic problems

Children may have scoliosis and or kyphosis. Hip problems could also be an issue for some children.

Gastrointestinal problems

Children may have a wide range of issues including constipation and diarrhea. Gastric reflux is common and increases the risk of aspiration.

Feeding issues

Children may be able to eat and chew typically but may have swallowing difficulties. This presents a risk for aspiration. A g-tube may help reduce aspiration and increase fluid intake. This is especially helpful during colds or other illnesses when eating and drinking may be more difficult.

Other reported findings that may be present but are not common

- Small hands

- Blood vessel malformations
- Pigmented areas of the skin
- Some evidence of an increased incidence of tumors most often choroid plexus papillomas
- Lower growth rate after ages 7-9
- Early or delayed puberty
- Difficulties regulating body temperature (extreme heat or cold)

What you can do

Encourage a comprehensive medical evaluation for the child each year. In addition to a primary care provider, the child will likely have many specialists involved with their care. Get copies of any changes to medications or care recommendations. It is important to meet with the parents to learn about the child's individual medical needs.

Seizures

- To minimize startle seizures: Give verbal warnings if anticipate loud noises (machines, vacuum cleaner, stapler, blender, etc.)
- Develop and implement seizure protocol for seizures at school
- If child has a Vagus Nerve Stimulator (VNS), those people supporting the child should know how to use the magnet to activate the VNS. This information should be part of the seizure protocol.

Respiratory

Monitor for any changes in cough, feeding difficulty, or energy levels and report promptly to parents. Follow any specific guidelines provided by parents and medical providers.

Gastrointestinal/Feeding

Follow guidance from parents and medical providers regarding diet and feeding protocols.

Education Supports

It is important to have high expectations for learning for children who have Aicardi, utilizing their strengths and interests as a foundation. Encourage use of the CORE educational curriculum and modify how it is taught to meet the child's individual needs.

What you need to know

- 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. Incorporate observations in child's natural settings as part of testing.
- Children vary widely in abilities, stamina, and medical challenges. Know each one individually and learn about their unique challenges and abilities.
- May have startle seizures in response to sudden sounds.

Vision

Vision abilities vary widely among the children who have Aicardi syndrome.

- Visual fields may be restricted and asymmetrical.
- The color combination black and white and the combination red and yellow were most often reported as the colors best seen by the child.
- Corrective glasses may be appropriate

Communication

- Communication skills range from children who are completely nonverbal, to those who have limited communication or use alternate forms of communication, and to those who have typical speech.
- Social skills are often a strength
- Continued communication support through the school years will be important as literacy and pragmatic capabilities (the use of language for social communication) become increasingly important in the middle and high school years.

Motor

- Many children who have Aicardi syndrome will have both fine and gross motor challenges.
- Some children will be able to walk, and some will use a wheelchair.
- Due to seizure activity, children who are mobile might need helmets to protect them if they fall during a seizure.
- Some children will have one side of the body that works better than the other side, which is important to know for motor activities and communication devices.

What you can do

Input from therapists and specialists may be needed to support classroom teachers and paraprofessionals. Obtain recommendations for medical providers and consider the following consults as needed to customize strategies to support the students' individual needs.

Physical and Occupational Therapy

- Physical therapy and occupational therapy may be needed for children with mobility and motor skills difficulties.
- Physical therapy may be helpful for children with orthopedic or strength issues.
- Occupational therapy can be helpful for problems with fine motor skills and to support

speech therapists in utilizing communication devices

Speech and Language Pathology/Communication

- Speech and Language therapy can help assist children with all aspects of verbal speech as well as with grammatical aspects of language both verbal and written.
- A speech-language pathologist may recommend use of an augmentative and alternative communication device (AAC) and/or the continued use of sign language to aid in expressive communication skills.
    - AAC are programmed for the individual child to provide them with a voice and ensure that the child can relay messages to others. (iPads are useful for many children.)
    - Find AAC system that allows for maximal social reciprocal communication.
    - Model use of the AAC device to encourage its use.
    - All persons interacting with the child should have education and training on how to encourage reciprocal communication with the device
- Consider multiple means of communication paired with the knowledge of when to use one method vs. another.
- Incorporate early use of augmentative communication aids such as picture cards or communication boards.
- Encourage finger pointing early to help with device use as they age.
- Communication should work with child’s desire to socially interact with others in natural settings.

Vision

If vision is affected, consult a vision specialist to understand the best visual field for communication devices. Corrective lenses may be helpful. Use high contrast color combination - black and white and the combination red and yellow were most often reported as the colors best seen by the child

Behavioral and Sensory Support

What you need to know

- Young children who have Aicardi syndrome often learn to respond to personal cues and interactions and can be very intuitive.
- Interest in other people allows children to express a broad range of feelings and form close bonds and real friendships.
- Children can and should be part of class activities, classroom jobs, and daily living skills.
- Children typically enjoy recreation, music, and physical activity.
- Crowds and loud noises may be difficult for some children, and may produce anxiety, fears, and/or worry.

What you can do

- Be proactive with behavioral supports. Discuss involvement of behavioral or mental health professionals, with the parents as needed.
- Make sure teaching strategies being used are appropriate for the child. For example, if the child is already socially engaged, make sure the interventions are suited for someone who is socially engaged.
- The use of firm and consistent directions, rules, and clear expectations are helpful.
- Consult OT to see if a sensory diet or other sensory supports may be helpful.
- Use positive behavioral interventions and supports
    - If there are behavior challenges, consult specialist to consider a functional behavioral assessment
- To minimize startle seizures: Give verbal warnings if anticipate loud noises (machines, vacuum cleaner, stapler, blender, etc.)
- Make sure children have opportunities for choice and control in their lives (choose colors, clothing, play, work partner, etc.

Physical Activity, Trips, Events

What you need to know

Changes in routine may produce anxiety, fears, and/or worry. Crowds and loud noise may be difficult for some children. Children with Aicardi Syndrome may have difficulty managing temperature so be conscious of the environment being visited.

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.

Bring appropriate layers, water, and any necessary medications. Monitor for evidence of overheating or chilling. Consider moving to a warmer or cooler location or taking a short break in a more temperate area for the comfort of the child.

School Absences and Fatigue

What you need to know

Absences may occur due to illness, medical appointments, seizure activity, or hospitalizations. Work with parents to minimize the disruption from absences. Difficulty sleeping may be an issue that can cause fatigue during the school day.

What you can do

- Discuss the child's nighttime sleeping patterns with the parents to determine if scheduling daytime naps or afternoon rests is needed.
- Provide consistent routine.
- Consider temperature in the environment and change to cooler or warmer if necessary.
- Some children respond to a change in scenery (i.e. taking a walk) when tired.

Emergency Planning

What you need to know

- Emergency plans will be individually determined, based on behaviors and medical issues.
- It is important to mention new signs, symptoms, or pain to the child's parents.
- Be aware of any temperature regulation issues that might be present and plan how to support the child as needed.

What you can do

- Review any health plans yearly or more often if the primary care provider sends updated recommendations
- Check to see if the child has a seizure protocol and know the plan in case there is a seizure.
- Have an ample supply of medications, food, formula, etc. for emergencies.
    - Check for expiration dates at least yearly and ask parents to replace any that are nearing their end date.

Resources

Medline Plus

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide good medical overview of the condition without being too technical.
https://medlineplus.gov/genetics/condition/aicardi-syndrome/

Aicardi Syndrome Foundation

The Aicardi Syndrome Foundation is a volunteer-based, non-profit organization dedicated to raising research funds and awareness for Aicardi syndrome.
https://aicardisyndromefoundation.org/

Our Aicardi Life

Meet other children with Aicardi and their families here. This is part of the Aicardi Syndrome Foundation's support network. http://ouraicardilife.org/

The Child Neurology Foundation

The Child Neurology Foundation serves as a collaborative center of education, resources, and support for children and their families living with neurologic conditions. They have a helpful entry about absent corpus callosum. See the useful resources under their “Tools and Resources” tab. https://www.childneurologyfoundation.org/disorder/agenesis-of-the-corpus-callosum/

Info for School Nurse and Primary Care Staff

Aicardi Syndrome is a complex condition that will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student's team. Work with the parents to get signed releases to share information at the school nurse's office and the primary care office.

Gene Reviews is a peer-reviewed point-of-care reference accessible via the National Library of Medicine. The entry for Aicardi Syndrome provides a comprehensive summary of the condition and describes some ongoing surveillance and management issues. It is a great resource for medical providers. https://www.ncbi.nlm.nih.gov/books/NBK1381/

HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, remind the parents to sign a release of information form at the primary care office. The school nurse should keep a copy of that release in the student's file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

Example letter for physician to provide to school

"My patient______________________ has been diagnosed with Aicardi Syndrome. This is a rare genetic condition that is characterized by multiple issues including: agenesis of the corpus callosum, visual problems, gastrointestinal issues, seizures, and developmental delays. This condition necessitates an Individualized Education Program (IEP) or 504 plan.

Physical therapy, occupational therapy, and speech pathology will likely be necessary parts of the child's education team. Please add their recommendations to my list of necessary accommodations. (*Include description of any visual deficits that will require specific supports and accommodations.)* Because of this condition, __________ needs the following modifications and accommodations:

Please inform teachers of the attached seizure management plan"

Created by NERGN with funding from HRSA grant UH7MC30778. Revised 5/2024

Meet a Child with Aicardi: *Jumping with Joy!*

"Joy is a sweet and loving girl" says her mother, Sherry. At age 5, Joy has so many friends in her day care and her school. She loves to meet and greet her friends and "the teacher thinks she even has a boyfriend!" quips Sherry. Sherry says that Joy is so excited every day when she goes to school that "she will run me over on the way to the door."

Joy was diagnosed with Aicardi syndrome when she was about 4 ½ months old. She spent several days in the hospital when her seizures began and the team arrived at the diagnosis during that hospital stay. At that point, her seizures were coming every hour and lasting about 30 minutes. Eventually, they shortened and got down to just a few minutes in length with the drug combination she was on. She also has had a vagus nerve stimulator (VNS) implanted to help with seizures.

Joy started school when she was three years old. She began walking shortly after at age 3 ½. Her day starts in a day care and then she is transported by the day care to the local school. She spends some of her day in a regular classroom and some in a special education classroom. She is able to feed herself with some assistance and can drink from a covered cup. They have found that if the water is chilled, she can drink it down with no problem. If it is at room temperature, she has a harder time and will choke. When she was younger, her mother discovered that Joy could hold her own bottle if it was a skinny bottle, so she bought cases of them at the dollar store!

Joy eats everything her family eats. Her food is chopped into small pieces so she can eat them easily. Her IEP lays out specifics around eating so she is safe at school. She also has some chairs that adapt as she grows to help her position while eating. Although she can walk, she may use a stroller after a seizure when her energy is low or when she needs a nap. Joy's hand is always held when she walks in case she has a seizure and might fall. They are hoping to get a seizure helmet soon as regular helmets aren't working.

Sherry thinks Joy can see fairly well while looking slightly to the left. They are still in the process of re-evaluating her vision.

Joy was speaking with single words and had as many as twelve words at one time. However, Sherry feels that when she came off some of the more effective seizure drugs (because they had reached the recommended length of usage), she began losing words as the seizures became less controlled. Recently, Joy started using two words again - 'Ma' and 'Oww' when she slipped off the couch and cried. She sometimes will slap to get people's attention. They are working on trying to get a more effective communication system in place.

Sherry's advice to other parents:

- Sherry feels that Joy teaches people what love is. She is very happy that she is included in her school and that her classmates have the opportunity to get to know her. All kids are a gift!

- Parents should go into meetings with a list and check off each item as it gets addressed. A prepared list helps when parents are feeling overwhelmed.

Sherry's advice to teachers, school nurses, and special education personnel:
Be patient with parents as they become acclimated to the school setting and meetings. Some have many fears and, as a team, you can all work through those fears together.

Thank you so much to Joy and family for sharing their story! And thank you to the team at the Institute on Disability at UNH for assisting in collecting these stories.