

Aicardi Syndrome *At a Glance*

Aicardi syndrome is a rare genetic condition found almost exclusively in girls. The complexity of this condition varies widely, with some girls having less obvious differences and others having more significant and apparent challenges. Girls with this condition typically have changes in their brain and eyes and have seizures.

The prevalence is estimated to be 1 in 105,000 – 1 in 167, 000 in the US.



Meet Joy on page 11

Things to Think About

1. Medical / Dietary Needs

What You Need to Know

Treatment involves managing 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

- Congestion
- Infections
- Obstructions
- Pneumonias
- These may be a result of aspiration

Orthopedic problems

- Scoliosis and or kyphosis
- Hip problems

Gastrointestinal problems

- Reflux
- Constipation
- Digestive issues

Feeding issues

- May be able to eat and chew typically
- May have swallowing difficulties
 - A g-tube may help reduce aspiration and increase fluid intake. It is especially helpful during colds/ illnesses when eating and drinking may be more difficult.

Physical characteristics and/or symptoms:

Not all people with Aicardi have all of these characteristics.

Aicardi was classically diagnosed by a triad of findings. As more individuals and families were identified, it became clear that there were additional major findings that helped make the diagnosis in individuals with only two of the triad. Therefore, there is evidence to suggest that two of the three findings in the triad (below), accompanied by at least two other major findings, may also indicate the diagnosis is appropriate.

Classic Triad of Findings:

- Absence of the corpus callosum (partial or complete)
 - Corpus callosum is the part of the brain which sits between the right and left sides of the brain and allows the right side to communicate with the left
- Infantile spasms
 - Many girls develop seizures prior to age 3 months and most by 1 year
 - Medically refractory (difficult to prevent) epilepsy with a variety of seizure types that develop over time

- Treatment often requires multiple anti-seizures drugs, ketogenic diet, or vagus nerve stimulator
- Eye findings
 - Lesions or lacunae of the retina of the eye
 - Lesions can be unilateral, bilateral, and asymmetric
 - Can have other eye problems
 - Some visual problems may lead to blindness

Other common findings:

- Other developmental brain abnormalities
- Underdevelopment of the optic nerve
- Low muscle tone in the trunk with increased muscle tone in the extremities
- Microcephaly (small brain), trunk hypotonia, and limb hypertonia with spasticity
- Moderate to significant developmental delay and intellectual delay
 - In some cases, mild to no learning disabilities are also reported
- Rib/vertebral defects are common
 - Can lead to scoliosis in up to 1/3 of affected individuals
 - Hip dysplasia
- Characteristic facial features
 - Short philtrum (space between nose and mouth)
 - Prominent upper jaw with upturned nasal tip and decreased angle of nasal bridge
 - Large ears, sparse lateral eyebrows
- Gastrointestinal difficulties
 - Parents report that these issues may be difficult to manage
 - Constipation
 - Gastro esophageal reflux
 - Diarrhea
 - Feeding difficulties. Child may have a feeding tube.

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)
- Some women in their 30's have this condition. However, life expectancy is variable and may be related to the severity of the seizures.

What you can do

Regular visits to doctors and specialists to help manage different challenges, as needed.

- Primary care doctor and care coordinator
- Ophthalmologist
- Orthopedist
- Gastro-intestinal doctor
- Neurologist
- Endocrinologist

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.

2. Education Supports

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

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.
- The girls (and occasional boy) vary widely in abilities, stamina, and medical challenges. Know each one individually and learn about their unique challenges and abilities.

Vision

Vision abilities vary widely among the girls 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.
- Light colors, pastels, and low contrast colors may be hardest to see.
- 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. A speech-language pathologist (SLP) may recommend use of an augmentative and alternative communication device and/or the continued use of sign language to aid in expressive communication skills.
 - These devices 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.
- An SLP can assist with speech clarity and rhythm of speech. An SLP can also assist with grammatical aspects of language in both spoken and written forms.
- 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 girls will be able to walk and some will use a wheelchair.
- Due to seizure activity, children who are mobile might need helmet 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

Communication

- A speech-language pathologist (SLP) may recommend the use of an augmentative and alternative communication device and/or the continued use of sign language to aid in expressive communication skills.
 - These devices are programmed for the individual child to provide them with a voice and ensure that the child can relay messages to others within functional settings such as the child's home and classroom.
- Speech and Language therapy can help assist children with all aspects of verbal speech including articulation, phonation, nasality, rhythm, and breathing as well as with language both verbal and written.
- 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.
- Incorporate typical language skills in social, work, and life skill areas.
- Children with speech and motor difficulties often benefit from speech and occupational therapy.
- Teach learning strategies for non-verbal communications.
 - Consider new technology, computers, and sign language.
 - Incorporate early use of augmentative communication aids such as picture cards or communication boards.
 - Communication should work with child's desire to socially interact with others in natural settings.
 - Make sure children have opportunities for choice and control in their lives (choose colors, clothing, play, work partner, etc.
 - Consider multiple means of communication paired with the knowledge of when to use one method vs. another.
 - Find AAC system that allows for maximal social reciprocal communication.
 - Encourage finger pointing early to help with device use as they age.
 - All persons interacting with the child should have education and training on how to encourage reciprocal communication with the device.
 - Model, model, model use of the AAC device to encourage its use.

- Continue with strategies that improve oral control to maximize their potential as oral speakers.
- Consider a team approach, such as an OT or PT to consultant to work with the SLP for optimal positioning to get the most benefit from hand use for communication.
- Consider vision specialists to gain insight into the best visual field for communication devices.

Consider therapists and specialists to consult and support classroom teachers and paraprofessionals

- Physical therapy
- Occupational therapy
- Speech therapy
- Vision therapy
- Musculoskeletal support
- Treatment for prevention of scoliosis related complications
- Alternative communication
- Paraprofessional support may be needed

3. Behavioral and Sensory Support

What you need to know

Social

- 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 family and class activities, household chores, and daily living skills.
- Children typically enjoy recreation, music, and physical activity.

What you can do

- 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*.
- Be proactive with behavioral supports. Discuss involvement of behavioral or mental health professionals, or medications with the parents as needed.
- The use of firm and consistent directions, rules, and clear expectations are helpful.
- Work with the occupational therapist to see if a sensory diet or other sensory supports may be helpful.
- Use positive behavioral interventions and supports
 - If there are behavior challenges, consider a functional behavioral assessment
 - A functional behavioral assessment, when used in the context of positive behavioral supports, is a method of developing an understanding of the function (purpose) of a person's challenging behavior and identifying positive ways to help the person have more effective and efficient ways of getting their needs met, preventing the occurrence of the behavior and changing other's responses so the behavior isn't reinforced.
 - Then develop a support plan:
 - Understand the causes of the behavior
 - Develop prevention strategies
 - Teach behaviors that WILL work to get what they want

Here are some resources related to Functional Behavior Assessments:

- Functional Behavior Assessment (from the Center for Effective Collaboration and Practice) <http://cecp.air.org/fba/>
- Functional Behavior Assessment (from the Least Restrictive Behavior Interventions Checklist) http://www.ideapartnership.org/documents/ASD-Collection/asd-dg_Brief_FBA.pdf
- Practical Functional Assessment Training Manual for School-Based Personnel (from Positive Behavioral Interventions and Supports) http://www.pbis.org/common/cms/files/pbisresources/practicalfba_trainingmanual.pdf
- Functional Assessment: What it is and How it Works (from Understood / for Learning and Attention Issues) <https://www.understood.org/en/school-learning/evaluations/evaluation-basics/functional-assessment-what-it-is-and-how-it-works>

- Resources / Practical Strategies for Teachers and Caregivers (from the Center on Social and Emotional Foundations for Early Learning)
<http://csefel.vanderbilt.edu/resources/strategies.html>
- Challenging Behavior (Technical Assistance Center for Social Emotional Intervention)
<http://challengingbehavior.fmhi.usf.edu/>

4. 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, or conversely, might be enjoyable.

What you can do

- Use preventative strategies
- 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

Absences may occur due to illness, medical appointments, seizure activity, or hospitalizations. Difficulty sleeping may be an issue.

What you can do

- Discuss the child's nighttime sleeping patterns with the parents.
- Provide consistent routine.
- Consider temperature in the environment and change to cooler or warmer 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.

6. 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

- 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.

7. Resources

GeneReviews: Aicardi

Learn more about the genetics of Aicardi Syndrome at:

<http://www.ncbi.nlm.nih.gov/books/NBK1381/>

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/>

Meet other children with Aicardi and their families on Our Aicardi Life, part of the Foundation's support network.

<http://ouraicardilife.org/>

NINDS Aicardi Syndrome Information Page

The National Institute of Neurological Disorders and Stroke (NINDS) offers their Aicardi Syndrome Information page at: <http://www.ninds.nih.gov/disorders/aicardi/aicardi.htm>

Classroom

Accommodations for Students with Visual Issues

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

Medline Plus

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

8. 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 and 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.