CONDITION

Aicardi syndrome

Learn about Aicardi syndrome, including infantile spasms, brain and eye changes, diagnosis, treatment, complications, and ongoing care.

WHAT IT IS
Rare brain and eye developmental condition

BODY SYSTEM
Nervous system and eyes

MAIN SYMPTOMS
Infantile spasms, developmental delay, vision problems, abnormal muscle tone

WHAT CAUSES IT
Unknown, suspected X-chromosome gene change

HOW IT IS DIAGNOSED
EEG, brain MRI, detailed eye examination

MAIN TREATMENT
Seizure control and supportive therapies

CAN IT BE CURED?
No, developmental changes cannot be corrected

IS IT AN EMERGENCY?
Sometimes, seizure lasting five minutes or longer

LONG-TERM OUTLOOK
Highly variable with possible shortened life expectancy

Overview

Aicardi syndrome is a rare condition that affects brain and eye development. It is usually recognized during infancy, often after a baby develops repeated seizures called infantile spasms.

The condition is classically associated with three findings: infantile spasms, incomplete development of the corpus callosum, and distinctive changes in the retina. The corpus callosum is the band of nerve fibers connecting the right and left sides of the brain. The retina is the light-sensitive tissue at the back of the eye.

The retinal changes are called chorioretinal lacunae. These are pale, rounded areas where layers at the back of the eye did not develop in the usual way. They are highly characteristic of Aicardi syndrome but do not always cause the same degree of vision loss.

Aicardi syndrome almost exclusively affects girls. Rare cases have occurred in boys with an extra X chromosome, usually an XXY chromosome pattern, or other unusual chromosome findings.

The condition varies greatly from one person to another. Some children have frequent seizures, major developmental disabilities, and significant feeding or movement problems. Others have better seizure control and develop more skills. Some people with Aicardi syndrome live into adulthood.

Early recognition matters because infantile spasms require prompt treatment. Ongoing care is usually coordinated by a pediatric neurologist and includes support for vision, movement, communication, nutrition, and learning.

Symptoms

Seizures are often the first noticeable symptom. Infantile spasms may look different from the shaking that many people associate with seizures. A baby may suddenly bend forward, stiffen, raise the arms, or draw up the knees. The movement may last only a second or two but repeat in clusters, especially after waking.

Spasms can be subtle. They may resemble a startle, brief head drop, or repeated crunching motion. A child may cry afterward or become less alert. Other seizure types can develop later, and seizures may arise from different areas of the brain.

Development is usually affected, although the degree varies. A child may take longer to gain head control, sit, move independently, communicate, or use the hands. Some children lose skills around the time seizures begin. Intellectual disability is common but ranges in severity.

Eye and vision findings can include:

A child's functional vision cannot always be predicted from the appearance of the retina. Brain differences may also affect how visual information is processed.

Muscle tone may be lower or higher than expected. Low tone can make head control and sitting difficult. High tone can cause stiffness, limited movement, or joints that become fixed in one position. Some children have weakness affecting one side more than the other.

Other possible features include difficulty feeding, slow growth, reflux, constipation, breathing problems, and sleep disturbance. Differences in the ribs or spine may cause scoliosis, an abnormal sideways curve of the spine. Not every person has all these symptoms.

- Chorioretinal lacunae
- Small eyes or other differences in eye development

- Gaps in eye structures called colobomas
- Abnormalities of the optic nerve, which carries visual information to the brain
- Reduced vision, unusual eye movements, or eyes that do not align

Causes and risk factors

The exact cause of Aicardi syndrome remains unknown. Researchers have long suspected that it results from a change involving a gene on the X chromosome that is important for early development. However, a single causative gene has not been confirmed.

Most cases appear to occur as new, or de novo, events during the formation of an egg or early embryo. This means the condition is generally not caused by anything a parent did before or during pregnancy.

Aicardi syndrome is not usually found in multiple members of the same family. The chance of it occurring again in a future pregnancy is believed to be low, but it cannot be considered zero. One reason is the possibility of germline mosaicism, in which a parent has a genetic change in some egg or sperm cells but not in blood cells used for testing.

There are no established lifestyle, environmental, or pregnancy-related risk factors. Parents who want information about recurrence can meet with a genetic counselor. Because the responsible gene has not been identified, counseling usually relies on the child's clinical findings, chromosome studies, family history, and available genetic test results.

Diagnosis

There is no single blood test that confirms Aicardi syndrome. Diagnosis is based on the pattern of seizures, brain imaging, and a detailed eye examination.

An electroencephalogram, or EEG, records the brain's electrical activity. In infantile spasms, an EEG may show a severely disorganized pattern called hypsarrhythmia. In Aicardi syndrome, abnormalities may be different on each side of the brain or may arise from several regions. EEG findings can change with age and treatment.

Magnetic resonance imaging, or MRI, provides detailed pictures of the brain. It may show that the corpus callosum is partly or completely absent. Other possible findings include fluid-filled cysts, enlarged brain spaces, unusually small or broad folds on the brain surface, and groups of nerve cells that did not move to their expected location during development.

An ophthalmologist examines the retina, optic nerve, and other eye structures, often after

using drops to widen the pupils. Identifying chorioretinal lacunae is especially helpful in distinguishing Aicardi syndrome from other conditions that cause seizures and an underdeveloped corpus callosum.

Additional testing may include chromosome analysis, chromosomal microarray, or broader genetic testing. These tests cannot currently rule Aicardi syndrome in or out, but they may identify another explanation for the symptoms. Chromosome testing is particularly important when the condition is considered in a boy.

The care team may also evaluate hearing, swallowing, nutrition, muscle tone, movement, and spine development. Because Aicardi syndrome is rare and variable, assessment by clinicians familiar with pediatric epilepsy and genetic conditions can be useful.

Treatment and management

There is no treatment that corrects the underlying developmental changes. Care focuses on controlling seizures, supporting development, maintaining nutrition and comfort, and identifying complications early.

Infantile spasms need prompt assessment because ongoing spasms and abnormal brain electrical activity may interfere with development. Treatments used for infantile spasms include hormonal therapy, such as adrenocorticotropic hormone or an oral corticosteroid, and the antiseizure medicine vigabatrin. The most appropriate option depends on the child's overall health, seizure pattern, prior treatment, and local specialist practice.

These treatments require medical monitoring. Hormonal therapy can affect blood pressure, blood sugar, infection risk, and other body systems. Vigabatrin can damage peripheral vision, although vision monitoring may be difficult when a child already has retinal or developmental abnormalities. Families should discuss expected benefits, important adverse effects, and the monitoring plan with the neurology team.

Many children develop additional seizure types and need more than one antiseizure medicine. Seizures associated with Aicardi syndrome can be difficult to control. If medicines are not effective enough, a specialized epilepsy center may consider options such as a ketogenic diet, vagus nerve stimulation, or epilepsy surgery. These approaches are not suitable for every child and require individualized evaluation.

Families should have a written seizure plan explaining what usual seizures look like, when to use prescribed rescue medicine, and when to seek emergency care. Antiseizure medicines should not be changed or stopped without guidance from the prescribing clinician.

Developmental care often begins in infancy. Physical therapy can support positioning,

movement, and flexibility. Occupational therapy can help with daily activities and adaptive equipment. Speech-language therapy may address communication and swallowing. Children who do not use speech may benefit from pictures, switches, eye-gaze systems, or other forms of augmentative communication.

Feeding support may include changes in food texture, seating, feeding techniques, or calorie intake. A swallowing study may be recommended if a child coughs, chokes, has frequent chest infections, or takes a long time to eat. Some children need tube feeding to provide adequate nutrition and reduce aspiration, which occurs when food or liquid enters the airway.

Vision specialists can assess usable vision and recommend high-contrast materials, lighting changes, or other adaptations. Orthopedic care may be needed for scoliosis, hip problems, joint stiffness, or difficulties with supportive seating. Treatment goals should reflect the child's comfort, abilities, and family priorities.

Complications

Frequent or prolonged seizures can lead to injuries, hospital care, or status epilepticus, which is a seizure emergency. Developmental disability may limit independent movement, communication, and self-care.

Swallowing problems can contribute to poor growth, dehydration, or aspiration pneumonia. Limited movement and abnormal muscle tone may lead to painful joints, reduced bone strength, hip displacement, or worsening scoliosis. Vision impairment can further affect movement and learning.

Respiratory illness and severe epilepsy may shorten life expectancy for some people, but outcomes are highly variable. Some individuals survive well into adulthood. A child's course cannot be predicted from one brain scan, eye examination, or early developmental assessment alone.

Follow up

Routine follow-up commonly involves neurology, primary care, ophthalmology, rehabilitation services, and developmental or educational teams. Some children also need gastroenterology, nutrition, pulmonology, orthopedics, or sleep medicine.

Appointments may review seizure frequency, medicine effects, growth, nutrition, bowel habits, sleep, breathing, mobility, pain, vision, and communication. Keeping a seizure diary or short video of unusual movements can help the clinician understand what is happening. Videos

should be recorded only when it is safe to do so.

Seek prompt medical advice for new clusters of possible spasms, a clear increase in seizures, loss of skills, repeated choking, poor intake, unexplained sleepiness, or new breathing symptoms. Suspected infantile spasms in a baby should be assessed urgently rather than waiting for a routine visit.

Call emergency services for a seizure lasting five minutes or longer unless the child's emergency plan gives different instructions. Emergency help is also needed for repeated seizures without recovery, severe breathing difficulty, blue or gray coloring, a serious injury, or failure to become responsive as expected after a seizure.

As children grow, follow-up should include planning for school support, equipment needs, puberty, transition to adult medical services, and future decision-making. Genetic counseling may be revisited if testing methods or knowledge about the condition change.

Sources

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