Aicardi Syndrome
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

Physical characteristics and/or symptoms

Note: not all people with Aicardi syndrome will have all of these features.

Classic Triad of Findings:

- Absence of the corpus callosum (partial or complete)
- Infantile spasms
- Eye findings
    - Lesions or lacunae of the retina of the eye

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, trunk hypotonia, and limb hypertonia with spasticity
- Moderate to significant developmental delay and intellectual delay
- Rib/vertebral defects are common
- Characteristic facial features
    - Short philtrum, prominent upper jaw with upturned nasal tip and decreased angle of nasal bridge.
    - Large ears, sparse lateral eyebrows
- Gastrointestinal difficulties
    - Constipation
    - Gastro esophageal reflux
    - Diarrhea
    - Feeding difficulties

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
- Lower growth rate after ages 7-9
- Early or delayed puberty
- Difficulties regulating body temperature

Recommended Routine Surveillance

- Routine dermatologic evaluation for vascular and other malignancies
- Monitoring and treatment of GI complications
- Regular monitoring of scoliosis

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

Emergencies should be handled as with any child.

- If seizures are present, the following seizure action plan may be useful:

https://www.aap.org/en-us/Documents/Seizure_Action_Plan_for%20School.pdf.

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Dermatology
  - Monitor for vascular and other malignancies
- Developmental evaluation
  - Physical therapist
  - Occupational therapist
  - Speech therapist
  - Vision therapist
  - Musculoskeletal support

- Endocrinologist
    - Precocious puberty or delayed puberty common
- Gastroenterologist
    - Monitor GI complications
    - Feeding tube might be necessary
- Geneticist / Genetic Counselor
    - Diagnosis
    - Coordination of care
    - Genetic risk for family
    - Clinical trials
- Neurologist
    - Management of seizures
- Ophthalmologist
    - Monitor eyes and vision
- Orthopedist
    - Scoliosis and vertebral defects
    - Wheelchair, stander or other medical equipment

Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

- My patient_____ has been diagnosed with Aicardi syndrome. Aicardi syndrome is a genetic condition that typically includes developmental brain abnormalities, seizures, and differences in the retina. Intellectual disability, low muscle tone or hypotonia, visual impairment, and gastrointestinal issues such as constipation, diarrhea, and gastroesophageal reflux are common medical issues. Some individuals with Aicardi have problems regulating their body temperature and many have skin changes that can include cancerous growths. Because of these, _____ needs the following accommodations.

Seven Important Aspects of School Life

"Aicardi at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events

- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

Gene Reviews
http://www.ncbi.nlm.nih.gov/books/NBK1381/

Genetic Home Reference
https://ghr.nlm.nih.gov/condition/aicardi-syndrome