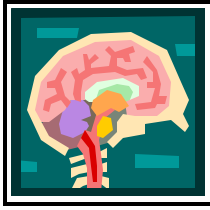

Disability Awareness Begins With You: Aicardi Syndrome

Definition

Aicardi Syndrome is a rare genetic disorder characterized by infantile spasms (jerking), absence of the corpus callosum (the connection between the two hemispheres of the brain), mental retardation, and lesions of the retina of the eye or optic nerve.

Causes, incidence, and risk factors

The cause of Aicardi Syndrome is unknown at this time, but is presumed to be a genetic disorder carried on the X-chromosome in some cases and a random, sporadic mutation in others. There are less than 500 cases of Aicardi worldwide.

Symptoms

Children are most commonly diagnosed with Aicardi Syndrome between the ages of three months and five months, if they meet the following criteria:

- Female sex (or XXY genotype male)
- Retinal lacunae -- lesions of the retina
- Seizures -- typically beginning as infantile spasms, a type of epileptic disorder in infants
- Absence of the corpus callosum (complete or partial)

The classic criteria remain the cornerstone of diagnosis in most patients. However, in rare cases, one of the features, especially lack of development of the corpus callosum, may be missing. The diagnosis can probably be made in such cases if two or more of the criteria below are present:

- abnormal formations in the brain, usually microgyria where the bumps on the brain are abnormally narrow
- Periventricular and subcortical heterotopia (a misplacement of groups of neurons in the brain sometimes found in seizure disorders)
- Cysts around the 3rd ventricle and/or choroid plexuses cysts in specific areas of the brain
- Papillomas of choroid plexuses (a benign, non-cancerous tumor found in a specific section of the brain)
- Optic disc/nerve coloboma (a congenital notch in the optic nerve)

Other Occasional findings:

- abnormal ribs or spine
- Microphthalmia -- a condition where eyes may be smaller than normal
- Other eye abnormalities, such as coloboma
- "Split-brain" EEG (dissociated suppression-burst tracing)
- Gross asymmetry -- large size differences between the two halves of the brain

Signs and tests

- An eye exam will be performed to confirm retinal involvement. Other occasional findings with the eye exam will be micropthalmia and coloboma. The degree or level of vision varies from normal to blindness.
- CT scan of the head or MRI of head can confirm lack of development of the corpus callosum, brain cysts, or other brain abnormalities.
- EEG can be used to help determine the type and severity of seizures.

Other procedures and tests may be performed depending on individual health concerns.

Treatment

Primarily involves medical management of seizures and any other health concerns, and early/ continuing intervention programs and therapies for developmental delays.

Support Groups

Aicardi Syndrome Newsletter Established in 1984 by amother of a daughter with Aicardi Syndrome in order to provide information and support for families due to the extremely rare nature of this disorder.

Publications include a bimonthly newsletter brief, and two to three large digests annually which are distributed to member families worldwide. The Newsletter has also published yearbooks, family directories, and two medical-model surveys of affected daughters.

Contact:
Denise Parsons, 1510 Polo Fields Court, Louisville, KY 40245, (502) 244-9152, E-mail: AICNews@aol.com

Aicardi Syndrome Foundation (ASF)

A not-for-profit, tax-exempt organization, incorporated in 1991. Working in cooperation with the Aicardi Syndrome Newsletter. The Foundation is funded by private donations and various fundraising events.

The A.S.F. has contributed financially to research into the causes of Aicardi Syndrome, and provides funds for the purchase of medical/ adaptive equipment for affected daughters, publication and distribution of the Newsletter, and biannual Aicardi Family International Conferences.

Contact:
Al and Cindy Meo, 450 Winterwood Drive, Roselle, IL 60172, 1-800-374-8518, E-mail: aicardi@aol.com

Additional information can be found at the Aicardi Syndrome web site: www.aicardisyndrome.com

Expectations (prognosis)

Prognosis varies widely depending upon the affected child's overall health and severity of symptoms. Nearly all have severe learning difficulties, although a few have some language abilities and some can walk independently or with support.

Many remain totally dependent for their care. The known age range of affected children is from birth to 30 years of age.

Complications

Complications vary according to the degree and severity of symptoms.

Calling your health care provider

Call your health care provider or seek emergency care for spasms in an infant or epileptic seizures.

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