Published on *My46* (https://www.my46.org) 01/20/2022

My46 Trait Profile

Aicardi syndrome

Aicardi syndrome is a rare genetic condition that affects mostly girls and is mainly characterized by structural brain problems, distinctive eye problems, and seizures. The gene associated with this condition has not yet been identified, but is believed to be located on the X chromosome.

Characteristics of Aicardi syndrome

Aicardi syndrome was initially characterized by a classic triad of agenesis of the corpus callosum (total or partial absence of the connection between the left and right halves of the brain), chorioretinal lacunae (punched-out appearing lesions of the retina – the light detecting layer at the back of the eye), and severe seizures called infantile spasms. It has since been found that individuals with Aicardi syndrome commonly have many other features. Almost all affected individuals have an abnormal location of brain cells (heterotopia), abnormal folds of the surface of the brain (polymicrogyria) and cysts inside the brain; some affected individuals have microcephaly (small heads). Additional eye problems include small or absent eyes, and abnormalities of the optic nerve. Individuals with this condition have moderate to severe developmental and intellectual disability. Their muscle tone can be reduced (axial hypotonia) or increased and stiff (hypertonia). Though not all individuals will experience infantile spasms, the majority will develop seizures before 1 year of age.

Aside from the neurological features, individuals with this condition may have abnormal development of the spine and ribs that can lead to scoliosis in up to 33% of affected individuals. The hands may be small and typical facial features, such as a short philtrum (the vertical indent between the nose and upper lip), slightly upturned nasal tip, large ears and sparse eyebrows have been reported. Additional features include constipation, diarrhea, gastroesophageal reflux, and feeding difficulties. Development of rare tumors has been reported in a few individuals. On average, affected individuals do not survive beyond childhood, but adults with Aicardi syndrome are known and the lifespan may be longer with improved management of complications.

Diagnosis/Testing

The diagnosis of Aicardi syndrome is currently made based on a clinician's assessment of an individual's signs and symptoms. An individual with all three of the features included in the classic triad could be given a diagnosis of Aicardi syndrome. The presence of two of the classic features and at least two other common features is also very suggestive of Aicardi syndrome. As we learn more about this highly complex condition, the diagnostic criteria may expand. The gene associated with this condition has not yet been identified but is believed to be located on the X chromosome.

Management/Surveillance

Management of this condition often involves a combination of occupational, physical, speech, and vision therapies. It is recommended that a pediatric neurologist be consulted, especially for seizure management which may include medications and vagus nerve stimulators (i.e., a device that is surgically inserted under the skin on the chest that sends electrical signals to the brain in an attempt to reduce the frequency of seizures). Some affected individuals develop large choroid plexus papillomas which are benign brain tumors that can lead to a build-up of fluid in the brain. Surgery may be necessary to remove such tumors. An ophthalmologist should also be consulted to look for lacunae or other abnormalities of the eye.

Imaging studies are recommended to evaluate for scoliosis or for other abnormalities of the spine and ribs. Because of the risk for vascular problems, routine dermatologic evaluations are recommended. Depending on symptoms, treatment for gastrointestinal complications may be warranted.

Mode of inheritance

The gene for Aicardi syndrome is believed to be on the X chromosome because most affected individuals are girls. It is thought that in males who typically have one X chromosome and one Y chromosome, Aicardi syndrome is fatal before birth because a second normal X chromosome is needed for survival to birth. At this time, the gene associated with Aicardi syndrome remains unknown.

Risk to family members

Aicardi syndrome is thought to be sporadic, and there are no confirmed cases of siblings with Aicardi syndrome, except for identical twins. Thus the chance of a couple having a second affected child is believed to be extremely low. Although there are some mildly affected individuals, there are no known reports of a mother passing on Aicardi syndrome to a child, and thus it is thought that all cases result from brand new (de novo) mutations.

Special considerations

None

Resources

Aicardi Syndrome Foundation
http://www.aicardisyndrome.org
Genetics Home Reference: Aicardi syndrome
http://ghr.nlm.nih.gov/condition/aicardi-syndrome
National Institute of Neurological Disorders and Stroke: Aicardi Syndrome
http://www.ninds.nih.gov/disorders/aicardi/aicardi.htm

References

Eble, TN. et al. (2009).Non-random X-chromosome inactivation in Aicardi Syndrome." Human Genetics 125(2): 211-216.
Fruhman, G. et al. (2012).Ophthalmologic findings in Aicardi syndrome." Journal of the American Association for Pediatric Ophthalmology and Strabismus 16(3): 238-241.
Glasmacher, MA. et al. (2007).Phenotype and management of Aicardi syndrome: new findings from a survey of 69 children." Journal of Child Neurology 22(2):176-184.
Kroner, BL. et al. (2008)."New incidence, prevalence, and survival of Aicardi syndrome from 408 cases." Journal of Child Neurology 23(5): 531-535.
Sutton, VR. et al. (2005).Facial and physical features of Aicardi syndrome: infants to teenagers." American Journal of Medical Genetics Part A. 138A(3): 254-258.
Sutton VR, Van den Veyver IB. (Updated 20 September 2012). Aicardi Syndrome. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at http://www.ncbi.nlm.nih.gov/books/NBK1381/. Accessed [08/16/2013].

Created: 08/2013 **Created by:** Tanya Eble, MS, CGC, Ignatia B. Van den Veyver, MD
Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC