Aniridia

Description

Aniridia is an eye disorder characterized by a complete or partial absence of the colored part of the eye (the iris). These iris abnormalities may cause the pupils to be abnormal or misshapen. Aniridia can cause reduction in the sharpness of vision (visual acuity) and increased sensitivity to light (photophobia).

People with aniridia can also have other eye problems. Increased pressure in the eye (glaucoma) typically appears in late childhood or early adolescence. Clouding of the lens of the eye (cataracts), occur in 50 percent to 85 percent of people with aniridia. In about 10 percent of affected people, the structures that carry information from the eyes to the brain (optic nerves) are underdeveloped. Individuals with aniridia may also have involuntary eye movements (nystagmus) or underdevelopment of the region at the back of the eye responsible for sharp central vision (foveal hypoplasia). Many of these eye problems contribute to progressive vision loss in affected individuals. The severity of symptoms is typically the same in both eyes.

Rarely, people with aniridia have behavioral problems, developmental delay, and problems detecting odors.

Frequency

Aniridia occurs in 1 in 50,000 to 100,000 newborns worldwide.

Causes

Aniridia is caused by mutations in the *PAX6* gene. The *PAX6* gene provides instructions for making a protein that is involved in the early development of the eyes, brain and spinal cord (central nervous system), and the pancreas. Within the brain, the PAX6 protein is involved in the development of a specialized group of brain cells that process smell (the olfactory bulb). The PAX6 protein attaches (binds) to specific regions of DNA and regulates the activity of other genes. On the basis of this role, the PAX6 protein is called a transcription factor. Following birth, the PAX6 protein regulates several genes that likely contribute to the maintenance of different eye structures.

Mutations in the *PAX6* gene result in the production of a nonfunctional PAX6 protein that is unable to bind to DNA and regulate the activity of other genes. A lack of functional PAX6 protein disrupts the formation of the eyes during embryonic

development.

Learn more about the gene associated with Aniridia

- PAX6

Inheritance

Aniridia is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In approximately two-thirds of cases, an affected person inherits the mutation from one affected parent. The remaining one-third of cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

Other Names for This Condition

- Absent iris
- Congenital aniridia
- Irideremia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Aniridia 1 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0344542/)
- Genetic Testing Registry: Congenital aniridia (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0003076/)

Genetic and Rare Diseases Information Center

- Isolated aniridia (https://rarediseases.info.nih.gov/diseases/5816/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Aniridia%22)

Catalog of Genes and Diseases from OMIM

- ANIRIDIA 1; AN1 (https://omim.org/entry/106210)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28aniridia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

References

- Chao LY, Huff V, Strong LC, Saunders GF. Mutation in the PAX6 gene in twentypatients with aniridia. Hum Mutat. 2000;15(4):332-9. doi:10.1002/(SICI)1098-1004(200004)15:43.0.CO;2-1. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10737978)
- Lee H, Khan R, O'Keefe M. Aniridia: current pathology and management. ActaOphthalmol. 2008 Nov;86(7):708-15. doi: 10.1111/j.1755-3768.2008.01427.x. Epub2008 Oct 6. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18937825)
- Robinson DO, Howarth RJ, Williamson KA, van Heyningen V, Beal SJ, Crolla JA. Genetic analysis of chromosome 11p13 and the PAX6 gene in a series of 125 casesreferred with aniridia. Am J Med Genet A. 2008 Mar 1;146A(5):558-69. doi:10.1002/ajmg.a.32209. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18241071)

Last updated June 1, 2009