

Aniridia

WHAT IS ANIRIDIA?

Aniridia is an eye disorder where the iris (colored ring structure of the eye that forms the pupil) is malformed. In some cases, other structures of the eye are also poorly developed. The word aniridia implies that there is “no iris,” but in fact there is a small ring of iris tissue present which is variable in size. Because the iris tissue is so small, the pupil is very large and may be irregularly shaped. Aniridia is a bilateral condition, meaning it is present in both eyes. However, the two eyes may be affected differently by the disease. [See Figure 1].



Fig. 1: Aniridia means an absence of the iris or colored part of the eye.

HOW COMMON IS ANIRIDIA?

In the general population, aniridia occurs in 1 per 50,000-100,000 people and the incidence varies in different regions.

WHAT CAUSES ANIRIDIA?

Aniridia occurs while the eye is developing during the 12th to 14th week of pregnancy. In most cases it is due to a mutation in the short arm of chromosome 11 (11p13) and affects the PAX6 gene, however it is also seen in genetic defects in nearby genes as well. The PAX6 gene provides instructions for making a protein that is involved in the early development of the eyes, brain, spinal cord, and pancreas.

WHAT IS THE CHANCE OF HAVING ANOTHER CHILD WITH ANIRIDIA?

Inheritance of aniridia can be familial (approximately 2/3 of cases) or sporadic (approximately 1/3 of cases). Familial aniridia is typically an autosomal dominant trait, so if one parent has aniridia, there is a 50% chance they will pass the gene on to the child of each pregnancy. When a child with aniridia is born to two parents who do not have the disease or the gene for aniridia, this is called a spontaneous mutation. A spontaneous mutation causing aniridia can occur either in the sperm, in the egg, or shortly after conception. The chance of the same parents having another child with aniridia is not any higher than in the general population since another spontaneous mutation would have to occur. A rare inheritance pattern of aniridia is autosomal recessive (when both parents carry the gene, but do not have the disease) in which a couple has a 25% chance of having a child with aniridia with each pregnancy. This type may be associated with intellectual disability.

HOW DOES ANIRIDIA AFFECT VISION?

There is a large range of visual outcomes in patients with aniridia. Patients may have very good vision (20/30 or better) to very poor vision (worse than 20/200), and most patients are somewhere in between. There are multiple components of the eye that may be effected in aniridia — not just the iris. Visual acuity depends largely on the extent of the disease and which components of the eye are affected.

Aniridia can be associated with abnormalities in the cornea (increasing risk of abrasions and causing blurred vision, glare, and scarring), in the angle structures where the cornea is adjacent to the iris (causing [glaucoma](#) in approximately 50% of patients), in the lens (causing [cataracts](#) in 50-80% of patients), in the optic nerve (e.g., hypoplasia), and in the retina (e.g., foveal hypoplasia). In addition, children often have some degree of [nyctagmus](#), which degrades visual quality, as well as large pupils, which cause light sensitivity.

Again, some children with aniridia have very good vision and have minimal associated ocular issues. It is difficult to predict what a baby with aniridia will see as they get older. The potential exists for many ocular complications from this disease, so it is important to have close and regular follow up with an ophthalmologist and sometimes with other ocular sub-specialists — such as cornea or glaucoma specialists — to ensure the best possible visual outcome is achieved.

IS ANIRIDIA ASSOCIATED WITH OTHER HEALTH PROBLEMS?



Aniridia may occur as an isolated finding or as part of a syndrome (i.e., a group of signs which occur consistently together). Miller syndrome is aniridia associated with a kidney tumor called a Wilms tumor (nephroblastoma). WAGR syndrome stands for Wilms tumor, Aniridia, Genitourinary abnormalities and mental Retardation. Gillespie syndrome is a combination of aniridia, mental retardation, and balance problems (ataxia). Patients with aniridia should strongly consider undergoing genetic testing and examination of family members. Sporadic cases of aniridia are much more likely to be associated with the development of Wilms tumor because there is more frequently a deletion of the PAX6 gene — unlike familial aniridia in which the gene is mutated but not deleted. Children found to have deletions of the PAX6 gene should have regular ultrasounds of their kidneys to screen for Wilms tumor. In addition, several systemic abnormalities have been associated with aniridia such as diabetes, metabolic disorders, obesity, autistic spectrum disorders, and neurologic problems, so a complete evaluation is warranted.

WHAT TREATMENT IS AVAILABLE FOR ANIRIDIA?

Children with aniridia need regular eye exams to assess vision and screen for glasses, glaucoma, and cataract. If glaucoma develops, treatment is typically with eye drops but may require surgery. If vision is reduced by a cataract, removal of the lens with or without an [intraocular lens implant](#) may be indicated. Lubrication of the eye surface with artificial tears and ointments can help with the associated corneal problems. Limbal stem cell transplants may improve corneal scarring. A recent development is a black, artificial iris-lens implant that is sometimes placed after cataract removal. Gene therapy clinical research is actively pursuing treatment options.

WHAT TYPE OF GLASSES SHOULD A CHILD WITH ANIRIDIA WEAR?

Your child's ophthalmologist will assess vision, examine them, and prescribe glasses if indicated. If no prescription is needed, patients often still benefit from wearing filter lens or sunglasses to help with glare and sensitivity to light (common symptoms associated with the large pupils and corneal changes). If a patient with aniridia also has low vision there are multiple services and visual aids available that may improve his or her functioning. Please ask your doctor for local sources.

WHAT ABOUT CONTACT LENS USE IN ANIRIDIA?

Some patients may be candidates for special, painted soft contact lenses, which can reduce glare, improve cosmetic appearance, and potentially improve vision. However,



extra caution should be exercised when using contact lenses if corneal abnormalities exist.

HOW OFTEN ARE EYE EXAMINATIONS REQUIRED?

Once aniridia is diagnosed, regular eye examinations for life are necessary. The frequency of examinations depends on what eye problems are present and is determined by the ophthalmologist. Frequent examinations will be needed in very young children and in children with severe disease. Additionally, abdominal ultrasounds monitoring for kidney tumors may occur as often as every 3 months with the primary care physician.

HOW WILL THE DIAGNOSIS OF ANIRIDIA AFFECT SCHOOLING?

It is important that a primary care provider assess a child's general development. Most children with aniridia attend normal school. The school's visual impairment team should be aware of a child's condition and maximize support in order to help continue mainstream education. Early intervention at school and at home makes a huge difference!

Are there any aniridia support groups?

- [Aniridia Foundation International](#)
- [Aniridia Network UK](#)
- [Vision for Tomorrow Foundation](#)
- [National Organization for Rare Disorders for information](#)

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