

Aniridia

WHAT IS ANIRIDIA?

Aniridia is an eye problem where the iris (colored part of the eye that is shaped like a donut and forms the pupil) does not develop normally. In some cases, other parts of the eye are also poorly developed. The word aniridia means that there is “no iris,” but in fact there is often a small ring of iris tissue present in eyes with aniridia. Because the iris tissue is so small, the pupil is very large and may have a funny shape. Aniridia affects both eyes, but, 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.

Fig. 1: Eye with aniridia showing the iris or colored part of the eye is missing.

HOW COMMON IS ANIRIDIA?

Aniridia is not very common. In the general population, aniridia occurs in 1 per 50,000-100,000 people and the number of people affected is different in different parts of the world.

WHAT CAUSES ANIRIDIA?

Aniridia happens while the eye is developing during the 12th to 14th week of pregnancy. In most cases it is due to a gene mutation. The most common mutation is in the short arm of chromosome 11 (11p13) and affects the PAX6 gene, however it is also seen when there are problems with other nearby



genes. The PAX6 gene gives instructions for making a protein that is very important in the early development of the eyes, brain, spinal cord, and pancreas. Aniridia can run in families (which happens in 2/3 of cases) or can be sporadic, meaning no one else in the family has the condition (which happens in 1/3 of cases).

WHAT IS THE CHANCE OF HAVING ANOTHER CHILD WITH ANIRIDIA?

When aniridia runs in families is typically as an autosomal dominant trait, this means if one parent has aniridia, there is a 50% chance they will pass the gene on to the child of each pregnancy and that child will get aniridia. 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 the two join together to form a baby. The chance of the same parents who do not have aniridia or carry the aniridia gene to have another child with aniridia is not any higher than the chance of getting aniridia in the general population since another spontaneous mutation would have to occur. A rare form 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 linked with brain disabilities.

HOW DOES ANIRIDIA AFFECT VISION?

The small or absent iris tissue in aniridia can cause light sensitivity and glare. However, in many cases, aniridia does not just affect the iris. Other parts of the eye can be affected as well in a way that can cause vision problems. Patients with aniridia can also have:

- Problems of the cornea (front clear shield covering the eye) – not enough stem cells, leading to corneal abrasions (scratches), glare, and corneal scarring
- Glaucoma – high pressure in the eye that damages the optic nerve- this occurs in about 50 % of patients
- Cataracts- cloudy spots in the lens of the eye- this occurs in 50-80% of patients



- Foveal hypoplasia – poor development of the part of the eye responsible for central, fine, detailed vision
- Nystagmus – this is a jerking movement of the eyes that can cause blurry vision.

Not all patients have all of these eye problems, There is a large range of vision 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.

It is difficult to predict what a baby with aniridia will see as they get older. It is possible to have different eye related complications from this disease, so it is important to have regular follow up with an ophthalmologist and sometimes with other eye specialists – such as cornea or glaucoma specialists – to help get the best possible vision.

IS ANIRIDIA SEEN WITH OTHER HEALTH PROBLEMS?

Aniridia may occur on its own as the only problem or as part of a syndrome (a group of health problems which are often seen together). Some rare syndromes with aniridia are:

- Miller syndrome - aniridia with a kidney tumor called Wilms tumor (nephroblastoma).
- WAGR syndrome - stands for Wilms tumor, Aniridia, Genitourinary abnormalities and mental Retardation.
- Gillespie syndrome - a combination of aniridia, mental retardation, and balance problems (ataxia).

Patients with aniridia should strongly consider getting genetic testing and examinations of other family members.

Sporadic cases of aniridia are much more likely to be seen with Wilms tumor because there is more frequently a missing PAX6 gene which leads to both problems. In familial aniridia, the gene is mutated but not missing (deleted). Children found to have deletions of the PAX6 gene should get ultrasounds of their kidneys to look for Wilms tumor. In addition, full medical check-up is recommended for patients with any form of aniridia as it can be seen with



diabetes, other metabolism problems, obesity, autism spectrum disorders, and brain (neurologic) problems.

WHAT TREATMENT IS AVAILABLE FOR ANIRIDIA?

Children with aniridia need regular eye exams to check for problems with vision, need for glasses, and look for signs of glaucoma and cataract. If glaucoma develops, there are different treatments including eye drops and surgery. If vision is blurry from a cataract, it may need treatment like glasses or surgery. Artificial tear drops and ointments can help with the corneal problems seen in aniridia. Stem cell transplants may help prevent corneal scarring from aniridia. An artificial iris-lens implant that is sometimes placed after cataract surgery can help with light sensitivity from aniridia. Gene therapy research is aimed at finding other possible aniridia treatments.

WHAT TYPE OF GLASSES SHOULD A CHILD WITH ANIRIDIA WEAR?

Your child's ophthalmologist will check vision and prescribe glasses if needed. If no prescription is needed, patients may benefit from wearing a special filter lens or sunglasses to help with glare and sensitivity to light from aniridia. If a patient with aniridia also has poor vision there are multiple services and visual aids that may help make the most of the vision. Please ask your ophthalmologist for more information.

WHAT ABOUT CONTACT LENS USE IN ANIRIDIA?

Some patients may benefit from using special, painted soft contact lenses, which can help with glare, improve how the eye looks, and may improve vision. However, it is important to be careful using contact lenses if there are already problems with the cornea.

HOW OFTEN ARE EYE EXAMS NEEDED FOR ANIRIDIA?

Once aniridia is diagnosed, it is important to get regular eye exams for life. How often the eye exams occur depends on what eye problems are present and is determined by the ophthalmologist. More frequent eye exams are needed in very young children and in children with severe disease. Additionally, abdominal ultrasounds looking for kidney tumors may be needed as often as every 3 months.



HOW WILL THE DIAGNOSIS OF ANIRIDIA AFFECT SCHOOLING?

It is important that a primary care provider assess a child's overall level of development. Most children with aniridia can go to a normal school. The school's team to help children with poor vision should be aware of a child's condition to help best support the child's 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](#)

For more scientific information on aniridia please see: <https://eyewiki.org/Aniridia>

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