ANIRIDIA FREQUENTLY ASKED QUESTIONS

1. Are people with aniridia blind?

No. Their vision may vary from 20/40 to 20/400. People with aniridia may be "legally blind" which is defined as vision that is not correctable to better than 20/200. However, this is quite different from total blindness. With 20/200 vision, one can still see colors, lights and the outline of large objects. In general, people with aniridia lack 'detail' vision and have better vision at close range.

2. What can someone with aniridia see?

The range of vision is different for each individual with aniridia, and there is not one specific description to explain what someone with aniridia can see. All people with aniridia will have challenges with seeing detail, due to the underdeveloped retina (foveal hypoplasia). Most pediatric ophthalmologists will tell parents of a child with aniridia "we will have to wait until he/she can tell us what he/she can see." This can be very scary and frustrating. The sight of a child with aniridia can develop and get better over time.

Generally, individuals with aniridia have a visual acuity measurement between 20/80 and 20/200. Some are legally blind, while others have vision good enough to drive a car. Most read without using Braille, especially in today's technically advanced environment of e-Readers and smart devices.

Some conditions related to aniridia are non-degenerative (do not get worse over time) and others are degenerative. Conditions that can degenerate the vision of

an individual with aniridia include corneal keratopathy, aniridic glaucoma, and cataracts. For more information, refer to the Aniridia's Impact on Vision section.

3. Will their vision get worse over time?

It can. The non-degenerative parts of this condition are the lack of iris (it will never grow) and the foveal hypoplasia (the underdeveloped retina). These conditions will not worsen with time (other than the "normal" changes that occur with age and are correctable with glasses). However, the impact of the secondary conditions such as glaucoma, corneal pannus and cataracts can worsen one's sight.

4. What causes the irregular eye movements?

This is a condition called nystagmus. The involuntary eye movements may be from side-to-side, up and down or rotary. Nystagmus is present to varying degrees in people with aniridia and typically declines with age. It tends to increase when the person is upset, excited or tired.

5. Does nystagmus affect vision?

Although people with nystagmus are not aware that their eyes are moving, it does make it more difficult for them to focus clearly on details. In fact, they will often find a "null point" which is the point where their nystagmus is the least apparent (see below).

6. What is the "null point"?

People with aniridia have a poorly developed fovea. In normal-sighted people, the fovea is in the center of the macula and an image will land there when the person is looking straight ahead. Their fovea is where they focus for their best detail vision – such as seeing a freckle on a person's face. Because a person with aniridia has an underdeveloped fovea, they must find their best area of focus which may be anywhere on their retina. Typically, they will move their head to the

position necessary to focus on this spot, consequently slowing their nystagmus and allowing for their best vision. This is their null point.

7. Is any of this correctable?

With the technology that is currently available, there are many treatment options for the secondary eye complications. First, there are instruments to measure eye pressure to keep glaucoma under control. If glaucoma is diagnosed, there are eye drops and/or surgeries to correct it. Also, there are surgical remedies for the corneal pannus such as stem cell transplants. Additionally, there are iris implants available to help decrease the amount of light that flows into the eye. However, this is not yet FDA approved. Please refer to the Aniridia's Impact on Vision section to learn more about secondary conditions and current treatments.

8. How does someone get aniridia?

In most cases, aniridia is a genetic condition caused by an anomaly in the 11p13 section of the PAX6 gene; however other genes have been identified in relation to aniridia. The genetic problem can be sporadic (happens for the first time within either the egg, sperm, or shortly after conception) or familial (passed on from one parent). Unlike most genetic conditions, this is autosomal dominant, meaning it takes only one mutated gene to cause this condition. Aniridia is not contagious. There is a fifty percent chance of an aniridic passing it on to one's offspring.

9. How can two people without aniridia produce a child with aniridia?

Both parents of a child with aniridia can be "normal," having NO genetic mutation or deletion of the PAX 6 (or other indicated aniridia) gene. If these two people have a child with aniridia, the aniridia is called "sporadic," The child obtained this genetic mutation or deletion from a spontaneous change in the egg, sperm, or soon following conception. The cause for this genetic mutation is not known. If these two parents should have another child, the chance of aniridia occurring in

the next pregnancy is the same chance as anyone else in the general population – very slim.

10. If an individual with aniridia has a child, what are the chances the newborn child will have aniridia?

Unlike most genetic conditions, aniridia is dominant. It only takes one parent to have the mutation or deletion to pass on this condition. Therefore, the chance of a man or woman with aniridia having a child with aniridia is fifty (50) percent. When someone with aniridia has a child with aniridia, the condition is called "familial" aniridia, because it was passed on by the parent with aniridia. With new technology, there is the possibility of identifying the mutation or deletion in the aniridia gene and using egg or sperm sorting to try to stop the aniridia from being passed on. This technology is very new and not much is known about its success rate.

11. How is aniridia diagnosed?

It is generally diagnosed by a pediatric ophthalmologist. In many instances, a pediatrician will notice during routine exam that a baby with aniridia does not have a pupil reaction to light. Or a parent may notice a large pupil area. A qualified ophthalmologist usually provides a diagnosis of aniridia during eye exams. In addition, a blood test performed by a geneticist can confirm the genetic mutation. For more information, contact a qualified genetic counselor. Genetic counselors are generally affiliated with universities and/or children's hospitals. Read more at https://visionfortomorrow.org/aniridia/aniridia-genetics-impact-on-vision/.

12. What is foveal hypoplasia?

The fovea is the part of the retina, in the center of the macula where one would normally focus for the sharpest most detailed vision. In people with aniridia, this

area is poorly formed or, in some cases, not present. This is known as foveal hypoplasia.

13. Is aniridia associated with mental retardation?

In most cases, aniridia is caused by a mutation in the PAX6 gene and is not associated with mental retardation. However, there is a small chance in sporadic (non-inherited) aniridia that it is caused by a missing, or deleted PAX6 gene, rather than a mutation. In these cases, deletion of other genes in the same area results in WAGR syndrome (WAGR is an acronym for Wilms tumor, Aniridia, Genitourinary abnormalities, and mental Retardation). Although mental retardation is common in individuals with WAGR syndrome, neither mental retardation nor any of the other conditions is always present. For this reason, genetic testing is recommended for all infants born with sporadic aniridia. For more information on WAGR syndrome: http://www.wagr.org/

14. Should an individual with aniridia have ultrasounds to test for Wilm's tumors?

Most pediatricians recommend that children with aniridia with a *deleted* PAX 6 be tested for Wilm's tumors every 3 months, as the possibility is high if there is a deletion. There is debate whether children with only a *mutated* PAX 6 gene undergo ultrasounds. Many pediatricians believe this non-invasive ultrasound is a safeguard for catching tumors early. Early detection is KEY in fighting these cancerous tumors. Therefore, the consensus is "it can't hurt" to get ultrasounds. As to how often, the safest procedure is to have the ultrasound every 3 months until the child is eight years of age when the probability of the Wilm's tumors decreases significantly. At that time, the monitoring is decreased to every 6 months or even annually. There has been a case of Wilm's tumors being detected in a 24-year-old individual with aniridia, so the annual monitoring is a good idea.

15. Do people with aniridia have to go to special schools?

No. Most children with aniridia function well in a mainstream classroom, with proper accommodations.

16. Are there special services for children with aniridia?

Yes. It's important for you to contact your county or state social service office to enroll your child in 'Early Intervention Services'. Each state has different agencies dedicated to ensuring all children needing early services receive them. Even if you feel your child is developmentally on track, it's important to be in contact with your local Early Intervention Services. The Center for Parent Information and Resources (http://www.parentcenterhub.org/find-your-center/)compiles information by state. Your pediatrician may be able to help connect you to the local agency, too, so don't hesitate to ask about it.

17. Can people with aniridia read books?

Yes! Although Braille is typically not necessary for people with aniridia, a few may learn to use it in order to give their eyes a rest. Depending on the degree of visual impairment, other accommodations may be necessary. These include large print books, a CCTV, magnifiers, and high contrast materials. e-Readers (like the Kindle and iPad) have made a significant impact on the ability to read any book because one simply has to adjust the font size to accommodate.

18. Do people with aniridia spend a lot of time going to doctors?

Yes! It is very important for aniridia patients to closely monitor each eye and to see the correct specialist for each problem (glaucoma ophthalmologists, cornea ophthalmologists, pediatric ophthalmologists, etc).

19. How does the sun affect the eyes of people with aniridia?

Most people with aniridia are sensitive to light. Imagine a normally-sighted person with their eyes dilated at an eye doctor appointment. This is how the eye

of someone with aniridia is at all times. Therefore, it is very important for them to wear sunglasses: a) for comfort and b) to protect the retina. One of the purposes of the iris is to protect the retina from too much sun. The retina can be damaged from too much sun exposure when an individual with aniridia does not wear sunglasses.

20. Can people with aniridia go outside?

Absolutely. With proper sunglasses, they can go anywhere, including the beach!

21. Can people with aniridia drive a car?

Sometimes. The requirements vary by country and by state. They may be required to wear a bioptic device to drive. For details, please see http://www.biopticdriving.org/

22. My child was just diagnosed with aniridia, what should I do?

Refer to our Newly Diagnosed – Aniridia section for more information.

23. What can I do to help my child 'make the most' of their functional vision?

Because your child's functional vision is dependent on many factors, it is important to get in touch with a qualified low-vision specialist to assess tools that may help your child. To find a low-vision specialist in your area, google 'low-vision specialist' + <your city>.

24. How can I find a qualified doctor in my area?

Aniridia is a rare condition. It is important that you find a doctor that has a base of patients with aniridia because some treatments that are common for some secondary conditions are contraindicated for individuals with aniridia. Ask your doctor how many patients he or she has with aniridia. If you are not comfortable or would like a second opinion, consider joining a social media group like Aniridic Family on Facebook, and post a comment asking for the type of specialist (e.g.

pediatric ophthalmologist, glaucoma specialist, cataract specialist, etc.) needed and in which area of the country you live. Many people with aniridia choose to travel far distances to receive the correct medical attention.

25. Sunglasses and hats are recommended throughout this website, can you recommend brands?

We find many families like the Julbo-brand (https://www.julbo.com/en/16/products/sunglasses/sfamily/junior_285.html) sunglasses in the 'Looping' style for babies because they are comfortable, the lenses are dark, and they are form-fitting which does not allow light to come in from the sides or above. Baby hats can be found in many retail establishments. We like Sunday Afternoons (http://sundayafternoons.com/) because they are lightweight, high-quality, and portable.

26. How can I meet others with aniridia?

You can start by contacting VFT's Family Support Coordinators Susie Chinn (susie.chinn@visionfortomorrow.org) and Debby Casher (debby@visionfortomorrow.org). Debby has been our Family Support Liaison for several years, and Susie joined the team in 2020. They are each raising a child with aniridia, and can offer suggestions for connecting with other families across the U.S. In addition, consider joining a social media group like Aniridic Family on Facebook.