Welcome

For some, your journey has just started, and others have a whole life full of experiences. Either way, you will be able to learn and share many things through Aniridia Foundation International.

In the past, there was little information and even less research about aniridia, mostly because ophthalmologists do not see many cases and are sometimes unfamiliar with its complexities. I felt the key to understanding this eye and medical condition called aniridia and to get researchers to make advancements, was to unite those affected by aniridia, their families, caring physicians, and determined researchers. Over the past eight years, Aniridia Foundation International (AFI) has made a real difference in the lives of many. We have made a difference by supporting families in the low vision community, creating awareness about aniridia, and spearheading important research. But we still have a ways to go.

The core of AFI is centered on research-based projects such as our largest and most important project, the International Aniridia Medical Registry and Gene Bank. United, we now have the tools (our collective medical data) and our AFI Medical and Scientific Advisors assisting us with our cause. By working together we can accomplish many things!

I was born with aniridia and glaucoma and my son inherited it, so I personally know the challenges of living with aniridia. You are not alone and we are here to help! We welcome you to join us and see how our efforts can one day Make a Miracle!

Jill Nerby
Executive Director,
Aniridia Foundation International

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www.aniridia.net
www.research4vision.com
About Aniridia Foundation International

Aniridia Foundation International (AFI) is a 501(c) 3 non-profit charitable organization dedicated to assisting those with low vision or blindness. Executive Director, Jill Nerby, founded AFI in 2001. Jill was born with aniridia and glaucoma and has undergone numerous surgeries to preserve her vision. Because of Jill's experience with this condition, as well as her passion to help others, educate the public, and a desire to make a difference for visually impaired and blind children of today and tomorrow, she invested her personal savings and time to launch this foundation.

In January of 2002, AFI board members Peter A. Netland, MD, PhD and Edward J. Holland, MD founded and currently co-chair AFI's Medical Advisory Board. Dr. Netland is a world renowned glaucoma specialist who spent several years as Director of Glaucoma Services at the Hamilton Eye Institute in Memphis before being chosen as Chair of the Ophthalmology Department at the University of Virginia where he presently resides. His research has focused on the pathophysiology of glaucoma and potential improvements in the diagnosis and treatment of this disease. Dr. Holland is Director of Cornea Services at the Cincinnati Eye Institute and Professor of Ophthalmology at University of Cincinnati, Cincinnati, Ohio. His expertise in corneal disease and cutting edge stem cell transplantation procedures have people from across the globe seeking his expertise to restore their vision. Dr. Netland and Dr. Holland now share AFI's Medical Board with 13 other national and International specialists.

AFI has experienced tremendous growth over its short eight years of existence. In 2002 AFI had approximately 25 families registered as members. Today, the organization has over 550 families as members. AFI is proud to report that an astounding 5,000 professionals, doctors and researchers are on the organization's medical newsletter circulation list and benefiting from AFI's impressive Medical Board. All this growth has taken AFI's mission to families from USA, Canada, Puerto Rico and over 15 other countries around the world.

Want to Help?

Volunteer on an AFI committee and become an active member.

Donations can be made by:

- Mailing a check to our address
- Making a donation online with a credit card at www.aniridia.net
- Enrolling in a United Way payroll deduction plan through your workplace
- Starting a corporate sponsorship (ask us for our corporate packet)
- Having your organization hold a FUNdraiser
Our Mission Statement

For many years, aniridia has been reported as somewhat rare, although we feel this condition may be more common than previously thought. If you only consider the aspect of being born with underdeveloped eye structures due to a genetic defect (retina, optic nerve, iris, etc), this in fact is uncommon. Aniridia is a multi-faceted condition comprised of eye and medical issues such as glaucoma, cataracts, corneal disease, diabetes, low vision and even blindness. To have many of these conditions in one person, such as in the case of a person with aniridia, it is uncommon. Yet these individual issues are very common in the general population. This is one reason our research goals often lead us to helping many others with low vision or blindness who share the same common issues as those with aniridia.

Ironically, even the naming of the condition many years ago is misleading. Named today, it would probably be called something else. Even though it’s the most noticeable physical characteristic, the term “aniridia” which means “no iris” is not entirely correct and is the very least of the problems for people with this condition.

Overall Goal

Aniridia Foundation International (AFI), along with its medical and research professionals seek to find a cure for aniridia. At the same time, through AFI's core research program, The International Aniridia Medical Registry and Gene Bank (IAMR), we seek to assist in the advancement of research and a cure of the additional conditions which make up the multi-faceted condition of aniridia. We believe that this AFI research program will continue to lead to improved understanding, better patient care, and lead us closer to a cure.

Immediate Goal

AFI believes that while research is important, helping infants born with aniridia and children / adults living their life with aniridia is also very important. Through educational and support programs and the AFI medical conferences, our goal is to assist in providing the information needed to allow them to make sound medical decisions about their own health care choices and provide the emotional support so that it will enable them to have a healthy, independent life.

Everyday Goal

Through our educational projects, medical meeting exhibitions and fundraisers, AFI seeks to promote awareness among the public and medical community and to dispel misconceptions about people with disabilities such as aniridia. Due to its low profile among other conditions, it is our goal to educate others about aniridia.

AFI was created to unite people with aniridia, their families, physicians, researchers and teachers to work together towards these goals.
Helping Other Eye and Medical Conditions Via Research

Being a subset of the low vision / blind community, and empathetic with others who live with the issues as we do, we have pledged to help advance research of aniridia and the eye and medical conditions which define it. The Aniridia Foundation International research program collects extensive medical data, DNA and works closely with several university research programs. We believe that through this research program we will help those with aniridia at the same time as others in the general population living with conditions we also experience such as glaucoma, corneal disease, diabetes, autism, etc. We seek patient support in participating in this research collection, as well as, research funding from supporters of the areas of which we may assist in advancing research.

Note:

Aniridia Foundation International (AFI) is a 501(c)3 non profit organization helping those born with low vision and blindness due to the genetic eye and medical condition aniridia. This condition is caused by a defect in the PAX 6 gene which is responsible for development of the eyes, kidney, pancreas, and the brain. Hard to control glaucoma, cataracts, corneal scarring / disease, low vision / blindness, retinal detachments, diabetes, obesity and autism spectrum disorders are all conditions found commonly amongst different people in the general population. The difference is that a child / adult with aniridia may be born or live with several of these conditions as there is no cure…yet. Please support AFI and Help Us Make a Miracle for those affected.

Executive Board

Jill A. Nerby
Executive Director

Peter A. Netland, MD, PhD
Chair, Dept. of Ophthalmology
Professor of Ophthalmology
University of Virginia

Christopher J. Albrecht, CPA
Albrecht CPA and Consulting

Edward J. Holland, MD
Director, Cornea Services
Professor of Ophthalmology
University of Cincinnati
Cincinnati Eye Institute

Medical Advisory Board

Co-Chairs:
Peter A. Netland, MD, PhD
Edward J. Holland, MD

Scientific Advisory Board

Co-Chairs:
James Lauderdale, PhD
Cellular Biology
University of Georgia

Ali Djalilian, MD
University of Illinois-Chicago
Member Benefits

Our organization was created to unite those who are born with the disabling eye condition, aniridia, and their families. Our international membership consists of those affected, their families, physicians, researchers, medical personnel, teachers and vision specialists.

Our member database serves many purposes: to keep members informed, to allow aniridia families to meet and share experiences, and to help the research community by providing medical and research data so we may find improved treatments and a cure.

We provide support, educational and social services to our members through many benefits and programs.

A free quarterly newsletter with various important information on aniridia, treatments, events, research, conference details, and regular columns like “Meet Our Member” and “Parents Pride”.

A website with medical, research and practical information for those affected, their families, and a special area for teachers / educators.

An online private “members only” area is also available. This area holds additional membership benefits for active AFI members who:

- wish to share information or post questions
- wish to participate in AFI special events such as the physician or researcher chat nights
- wish to get discounted rates, advance registration, special event invites, and other perks for conferences, events, products.
- wish to develop friendships and support networks.

Please see membership folder for details.

“Be a part of the solution.”

Become an active member by volunteering or donating to AFI. If you are not an active member yet, go to our website at www.aniridia.net and fill out member form online, or download the form and mail or fax it to us.

“I felt an instant support system.”

Page Ferrell, diagnosed with aniridia

“It was invaluable to meet other families coping with aniridia.”

Will Richardson, son of Victoria & Craig Richardson, diagnosed with aniridia
Aniridia Foundation International (AFI) seeks to use its funds wisely and to get the most out of every dollar. Therefore, we try to create programs that have dual purposes to make the most of our funds. As you will see, the beauty of these programs, being complimentary to each other, is that AFI can serve the needs of research and patient care at the same time.

Research
Lauderdale Laboratory:
Dr. James Lauderdale, PhD is Co-Chair of the Aniridia Foundation International Scientific Advisory Board and helps AFI with their research programs such as the International Aniridia Medical Registry and Gene Bank.

In the Lauderdale Laboratory at the University of Georgia, researchers study the role and function of the PAX6 gene in the human eye. This gene is required for proper development of the eye prior to birth and is involved in maintaining the eye throughout childhood and adulthood. Lauderdale Laboratory is involved in exciting research that could someday be used to treat defects of the eye and help to restore vision in individuals who suffer from PAX6-related disorders. Aniridia Foundation International is a substantial contributor to the Lauderdale Laboratory.

International Aniridia Medical Registry and Gene Bank:
We have created an International Aniridia Medical Registry where we are currently collecting data on aniridia and its associated or suspected associated conditions from all people born with aniridia. Our gene bank will help with research and education of the medical community through clinical photographs, DNA and tissue samples. Both aniridia and glaucoma will be studied with the data through the registry and gene bank. Our current research goal is to obtain enough funding so that we may help fund new aniridia research and its conditions that affect people with aniridia such as: glaucoma, corneal scarring, nystagmus, cataracts, and the genetics involved in each.

CUDDLES and WE CARE Programs
These programs focus on the emotional support for children (Cuddles) and teens and adults (We Care). In Cuddles, stuffed teddy bears or animals with personalized t-shirts are sent to infants and children who are either going through surgery, transplants, chemo-therapy or dealing with stressful situations. These Cuddles bears have been used in play therapy to help children overcome the fear of eye examinations or surgery when parents and/or the doctor examine the bears’ eyes first or practices preparations for surgery with the bear. Teens and adults may also experience fear, need someone to talk to, or just feel good that people are thinking of them during this time. In the We Care program, when a person is referred, we send out an age appropriate “thinking of you” item.

(continued)
Education

Awareness in the Medical Community:
Each year, Aniridia Foundation International exhibits at several medical conferences put on by organizations such as the American Academy of Ophthalmology (AAO), American Academy of Pediatrics (AAP), World Cornea Congress (WCC), American Society of Cataract and Refractive Surgery (ASCRS), and the Eye Bank Association of America. We also have future plans of exhibiting at low vision and National Education Association meetings.

Our presence at these distinguished medical conferences has raised the awareness of aniridia and created interest in research opportunities.

AFI Medical Conference:
Through AFI's biannual medical conferences and with the help of our Medical Advisory and Scientific Boards, we bring in top physicians and researchers to speak on aniridia and its associated conditions, the latest research, new treatments and topics to enrich the lives of those with aniridia and their families. Since aniridia is a subset of the low vision and blind population, we also have seminars on general low vision and blind topics to help people maintain a quality of life. This is also a time when families can get know others with aniridia and make special connections with people who can relate to their personal experiences.

The HOPE Fund
This fund's primary focus is to help offset costs for needy families or individuals who want to attend AFI's medical conferences. AFI can give these families the opportunity to have the education they need from our renowned doctors and researchers to make good medical decisions. Our medical conferences have consistently been valued highly among those who have attended as a great time to connect with others. Individuals or families must furnish financial proof of need and will be selected as funds allow.

OPTIC Program
This program has helped many individuals with aniridia and other cornea patients who experience the corneal scarring that those with aniridia do. Aniridia Foundation International is proud of its involvement in helping to create this program. In the beginning, the majority of the OPTIC patients were those with aniridia. During that time, AFI granted over $70,000 since the inception of this program and is open to further partial funding requests in the future.

Those that have benefitted are those with cornea problems such as aniridia, Stevens-Johnson Syndrome, chemical and thermal burns to the eyes, extensive contact wear patients and other corneal diseases.

This program is to assist those in these conditions through a procedure called the Keratolimbal allograft or KLAL. In this procedure, stem cells from cadaver donors are transplanted onto eyes of those blinded by certain corneal diseases. Close monitoring after a KLAL stem cell transplant is very important in retaining sight. It is also important for patients to stay educated about their medications and health after the procedure. The OPTIC program is made up of a team of doctors and an OPTIC coordinator. The team of doctors consists of one corneal, retinal, glaucoma, ocuplastic and nephrology/transplantation specialist. The OPTIC Coordinator is a Registered Nurse experienced in transplantation and the associated medications.
The coordinator is the liaison between the medical team and the patient. The coordinator educates the patient of the compliance issues required for the KLAL procedure. Blood work is monitored on a monthly and quarterly basis with the results reported to the team of doctors. This doctor-patient interaction is paramount in managing the health of the patient and minimizing any side effects from the medications. The beauty of these programs being complimentary to each other is that AFI can serve the needs of research and patient care at the same time.

Website
Our public website gives basic information on aniridia and its associated conditions such as glaucoma, corneal scarring, cataracts, low vision, Wilms’ tumor (a cancerous tumor of the kidney in young children) and diabetes.

Members and the Public:
For our members, we have an area where members can meet and connect with others, ask questions, discuss topics, gain support, and develop friendships.

Physicians and Researchers:
We also have a special area for physicians and researchers entitled the AFI Knowledge Transfer Center at www.pax6.com.

Newsletters

Members and the Public:
AFI’s quarterly newsletter for members and the public is called “Eye on Aniridia” and through generous donations, are free, which allows members at every economic level to be educated, supported and feel like they belong. News on aniridia and its associated conditions, research and opportunities to participate, helpful aids and tips, conferences and fundraisers, are all included. Some of the regular columns are “Meet Our Member”, “Parents Pride”, and “From the Director.”

The newsletter it is available in large print, audio (CD), and in PDF / Word formats for those with low vision or blind using an enlarging or screen reader computer program.

Physicians and Researchers:
AFI’s semi-annual newsletter for physicians and researchers is called “Aniridia InSight” and is distributed via mail and email to these professionals around the world. The articles are written by medical and research professionals for those desiring to learn more about aniridia, its associated conditions, treatments and research. This is an excellent way to keep the clinical community and the research community aware of the latest developments and ways they can help each other.
What is Aniridia?

Description
Aniridia is a genetic eye condition usually detected at birth. Its approximate occurrence rate is 1/50,000 births. Aniridia is produced by a failure in development of the ocular globe during pregnancy due to a genetic mutation, a deletion in the short arm of the chromosome 11p13 affecting the PAX6 gene, which is responsible for the formation of the eye.

The term “aniridia” means without iris (the colored part of the eye which expands and contracts to control light), and although not entirely absent, all that remains of the iris is a thick collar of tissue around the outer edge. The muscles which open and close the pupil are missing entirely. In addition to the lack of iris, many people with aniridia have other severe eye conditions which further limit vision. Such associated problems are: glaucoma, malformation of the retina, corneal degeneration, amblyopia, foveal hypoplasia, cataracts, nystagmus, lens subluxation (dislocation), and macula and optic nerve disease. Most people with aniridia have vision of 20/200 or worse and are considered legally blind.

Since aniridia is a genetic disorder, there are other conditions that are sometimes associated with it such as WAGR Syndrome. WAGR is an acrostic for Wilms’ tumor (cancerous tumor of the kidney usually occurring before age 8 years old), Aniridia, Genitourinary abnormalities or Gonadblastoma, and Retardation. This larger genetic deletion includes the PAX6 gene and the nearby WT1 gene. About one-third of those born with sporadic cases of aniridia will have the WAGR Syndrome.

Associated Conditions
Glaucoma: Glaucoma is called the “thief of sight” because there may not be any signs before much damage is done. Since glaucoma in people with aniridia can develop at any age including birth, it is important to have the child’s pressure checked at birth and every 6 months thereafter.

Wilms’ Tumor: Most pediatricians suggest genetic testing if a diagnosis of sporadic aniridia is found at or shortly after birth. This is because of the possibility of WAGR Syndrome.
Due to some unusual findings of normal genetic karotype patients later producing a Wilms’ tumor, we suggest that all children up to eight years old with sporadic aniridia have ultrasounds every 3 months to monitor their kidneys. While the risk is lower with a genetic test in which the WT1 gene is intact, this ultrasound monitoring is simple, non-invasive, and is a good preventive measure. Most Wilms’ tumors, if caught early, can be cured.

**Corneal Scarring (Degeneration):** People with aniridia usually develop corneal scarring at some point in their life. Most are seen in their teens and 20’s. However, it can develop earlier, especially if there is a trauma to the corneal surface such as a scratch or an abrasion from a contact lens. It has been estimated that about 90 percent of those with aniridia develop the corneal scarring. The cornea is the outer layer of the eye in front of the iris and pupil. The reason for this scarring is due to the eye’s limbal stem cells either being underdeveloped or not working properly to generate new corneal tissue. Ironically, these limbal stem cells are located in the area where the underdeveloped iris is located.

These cells are responsible for making new cornea cells to keep the corneal surface healthy and clear. However, the limbal stem cells in a person with aniridia do not work well. When their cornea cells are sloughed off, the cells from the white part of the eye (sclera) grow in to cover the area. This Condition is called Limbal Stem Cell Deficiency (LSCD). The cells from the scleral area are not transparent like the cornea cells. This is what reduces the vision of the person with aniridia.

Limbal stem cell deficiency can be found early by a corneal specialist experienced in aniridia. As it occurs, reading becomes difficult, then larger objects or people are difficult to see. As the condition worsens, the person will only see light and dark contrasts. The good news is that LSCD can be treated with good results if caught early. We suggest to all our members to have their children monitored so that LSCD can be treated when it is necessary by an experienced corneal surgeon who is knowledgeable about aniridia. It is also important to note that corneal transplants are not successful for those with aniridia without a limbal stem cell transplant being done first.
A Guide for Parents

We know that for some of you, the diagnosis of aniridia has come as a complete shock. But we are here to help you learn, be supportive, and provide advice. We have members young and old who were born with aniridia and are willing to share their experiences. Parents and families are also involved and can provide support from their perspective.

Our Medical Advisory Board spans many different specialties of doctors and researchers. They have been chosen because of their experience in treating people with aniridia, or their work in aniridia research and their strong dedication to helping us.

Will My Baby See?

Your baby was born with eye structures that are underdeveloped due to a genetic error. The optic nerve, retina, lens, and iris can all be affected and may cause different visual acuities from child to child depending on the extent of underdevelopment. The lack of a fully developed iris will be the most noticeable feature, yet the least of concern. It may look like your baby has dark eyes, however, it is like an enlarged pupil surrounded by the iris stump that never grew. Some with aniridia will most likely experience some sensitivity to light, a medical condition known as photophobia.

The absence of iris doesn’t cause blindness; however, proper monitoring by an ophthalmologist who has experience with aniridia will watch for the associated conditions such as glaucoma, cataracts, strabismus, nystagmus and corneal scarring which can limit vision or cause loss of vision.

Most young children have a visual acuity of between 20/80 and 20/200. The ideal vision of a person is 20/20. A baby’s visual acuity can be estimated early in life by how they react to parents faces, their interest in objects and the ability to follow objects from side to side. Since infants with aniridia have some vision, they should learn to walk and develop as a child appropriate for their age. However, there may be depth perception difficulties. This may cause some problems with hand-eye coordination. For this reason, kids may seem clumsy and should be careful when walking around changes in the ground level and near steps. Fortunately, there are many adaptations that can help.
What is Photophobia?

Think of the eye as a camera and the shutter is the iris. If too much light is let into the camera lens, the picture will be washed out or overdeveloped. This makes the definition of objects hard to see. When too much light falls onto the retina, it can cause discomfort and loss of visual acuity. Artificial lighting indoors can also cause glare.

Some children may try to control light by squinting or have a condition known as ptosis (toe-sis). This is where the eyelids droop to compensate for the light entering the eye. Often it may look like the child is sleepy to others who do not know of the eye condition.

Vision can be diminished for anyone when glare is involved. For this reason, pick sunglasses for babies and children with polarized lenses to block out the glare. There are special sunglasses made for babies and a brimmed hat can also help.

Why do Doctors want Genetic Tests?

Aniridia is produced by a failure in development of the ocular globe during pregnancy due to a genetic mutation, a deletion in the short arm of the chromosome 11p13 affecting the PAX6 gene, which is responsible for the formation of the eye. About 2/3 of all cases are inherited (familial), but it can also happen sporadically (neither parent has it). The error can occur in the father’s sperm, in the mother’s egg, or after conception. Aniridia is an autosomal dominant condition which means it only takes one defective PAX6 gene to produce this eye condition.

When the parent has aniridia and passes it on to their child it is known as Familial aniridia. When the person with aniridia is involved with a pregnancy, there is a 50 percent chance with each pregnancy, that the child born will also have aniridia. In Aniridia Foundation International there are many families who have several generations of members with aniridia.

Sporadic aniridia happens in the other 1/3 of aniridia cases. Of the sporadic cases, 1/3 of those will have a more severe disorder called WAGR syndrome. This is caused by a larger deletion of the 11th chromosome involving the PAX6 gene and the nearby WT1 gene, responsible for the Wilms’ tumor. WAGR syndrome stands for Wilms’ tumor (cancerous tumor of the kidney usually occurring before age 8 years old), Aniridia, Genitourinary abnormalities or Gonadblastoma, and Retardation. To be diagnosed with WAGR, a child will have aniridia and at least one of the other symptoms of WAGR.

This is why doctors want to have genetic tests. It will give them an idea of what to watch out for medically and developmentally so that your
child can get the best possible care and monitoring.

Doctors suggest that all children up to eight years old with sporadic aniridia have ultrasounds every 3 months to monitor their kidneys. There have been reports of older people getting Wilm's tumor; however, it is rare. Many parents move to a longer interval between ultrasounds after age eight, after consulting their doctor.

Early Intervention is Important

When your baby is diagnosed with this low vision eye disorder, you should contact your Department of Human Services to inquire about early intervention options and an assessment. Therapists will work with your baby to help him or her develop to his or her best potential. There are many moms and dads in Aniridia Foundation International that can help you with information and support you during this difficult time.

A Message to Remember

It may seem overwhelming now; however, our foundation gives you the tools to learn through our medical conferences where we bring in top doctors experienced in aniridia to speak to you, and support networking with families who have experience with the challenges of aniridia. You will meet many children, teens and adults with aniridia who have excelled despite their limited vision and can help you understand what your child's life might be like. Grandparents, parents and other relatives are also members and can give their very special perspective and support.

“We discovered we are not alone in this journey.”

Mark & Heidi Petre with daughter, Anna, diagnosed with aniridia

Take Our Hands, Walk with Us, Share Our Dreams, and Help Us Make a Miracle!

Aniridia Foundation International
Our Book

We are very happy to report our first book, *Aniridia and WAGR Syndrome: A Guide for Patients and Families* was released Spring 2010! Additionally, we are pleased to share that due to a special agreement AFI worked out with the publisher, all books will come with a copyrighted CD. We wanted the book to come on CD so if you have a screen reader or zoomtext program it would be easier for you to read it.

To order, please fill out the form below, or for international orders use:

Oxford Publishing (UK and Canada)
Amazon.com (UK, Ireland, Canada, Germany, Austria, France and Japan)

Aniridia Book Order Form

We are pleased to offer our first book, *Aniridia and WAGR Syndrome: A Guide for Patients and Families*. Included with the book is a CD version (as a Word document for Screenreader and as a PDF for ZoomText).


Number of Books ______ x $35.00 = ________

Shipping:
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