



Excerpted from “**Just Diagnosed: A Guide for Parents**”
Courtesy of the Aniridia Network

What is Aniridia?

Aniridia is an eye condition named after its most obvious symptom. Aniridia literally means "without irises" (the colored part of the eye) People with Aniridia have an enlarged pupil, so their eyes appear black. Aniridia is a developmental disorder; the eye stops growing properly early in pregnancy, which can cause problems in the eye later in life.

The following conditions may be present at birth, or may develop later in life:

Glaucoma (increased pressure within the eye).

People with Aniridia have a particularly high risk of developing glaucoma, and it may be very difficult to treat. Regular testing should begin at birth.

Cataracts (areas of clouding in the lens of the eye) Often present at birth. If cataracts interfere with vision, they may require surgical removal.

Macular or Foveal Hypoplasia (underdevelopment of the fovea or macular region of the retina) Macular hypoplasia is a frequent cause of poor vision.

Aniridic keratopathy (also called “corneal pannus,” it is a clouding of the cornea of the eye caused by an overgrowth of blood vessels). Keratopathy may be noted in childhood or adolescence, but typically progresses between the ages of 20 and 40. The risk of this problem is increased by eye surgery.

Can My Baby See?

Lack of an iris alone does not cause blindness. Functional vision in an individual will depend on the type and degree of other conditions, such as the development

of the macula, the presence of cataracts, or the degree of glaucoma. The amount of functional vision can vary dramatically between individuals, but most people with Aniridia have visual acuities in the 20/80 to 20/200 range.

Visual Acuity, (the ability to see) is measured in the following way:

20/20 = what the average person can see at 20 feet, the individual being tested sees at 20 feet.

20/80 = what the average person can see at 80 feet, the individual being tested can only see at 20 feet.

20/200 = what the average person can see at 200 feet the individual being tested can only see at 20 feet.

Whether a baby can see can usually be determined fairly early in life by their interest in lights, objects, and faces, and their willingness to follow these objects of interest from side to side. Facial expression also suggests active vision.

But while parents of typically-sighted children usually note these early signs of vision in infants 2 weeks to 3 months of age, it is not unusual for babies with aniridia to begin to display them at a much later age. Parents of children with aniridia often say that their child seemed to "wake up" visually as late as 6 to 9 months of age.

Because it is likely that your child will have some vision, it's important to provide many opportunities for them to use the vision they have. There are many books and resources to assist parents with activities which will encourage their child with low vision to develop to their full potential.

What is Photophobia? How Can I Help My Child With This Problem?

Photophobia is common in people with aniridia. Photophobia literally means "fear of light." It is not a true phobia, but a medical condition in which a person feels discomfort or pain in the eye when bright light is present. People with aniridia may experience this discomfort in sunlight or indoors.

In the normal eye, the irises provide protection against the sun. Since studies have shown that too much sunlight on the retina may increase the risk for cataracts in all humans, people with aniridia must be particularly careful to protect their eyes when outside.

Small children with aniridia seem to have an especially difficult problem with sensitivity to light. This difficulty seems to lessen as they grow older, perhaps because they learn other ways to control the amount of light that enters the eye by squinting.

Artificial (indoor) light is not harmful to people with aniridia, and is usually not bright enough to be uncomfortable, so it is not necessary to darken indoor lights for a child with aniridia. When out of doors - even on cloudy days - dark sunglasses and a hat are important for both protection from ultraviolet rays and for comfort and safety.

Finding and fitting infants and young children with sunglasses can be a real challenge. An excellent resource is <http://www.babysunglasses.org>. This website is maintained by the mother of a child with aniridia, and has an excellent review of many styles of sunglasses, with photos, prices and ordering information from a wide variety of manufacturers.

What about Glasses?

One of the first questions families ask about their child with aniridia is whether prescription glasses would be helpful for them. To answer this question, it is important to understand how prescription lenses work. Glasses correct vision when the eye is "near-sighted" or "farsighted" (or a combination of these). These problems are the result of the front of the eye being slightly shorter, longer, or more irregularly shaped than is normal. This irregular shape distorts light as it comes into the eye, and causes blurred vision. Placing a lens in front of the eye corrects the distortion, and allows vision to be clear.

Some people with aniridia are near-sighted or far-sighted, and glasses can improve that aspect of their vision. However, most people with aniridia have decreased vision as a result of abnormalities at the back of the eye, in the fovea or macula. These abnormalities cannot be corrected with glasses.

Contact Lenses

In years past, special contact lenses were often prescribed for people with aniridia. These contact lenses had an artificial iris painted on them, and were thought to help decrease glare. In more recent times, dark tinted contact lenses have been used for the same reason. New research on aniridia shows that the aniridic cornea lacks certain important cells, called "limbal cells," which allow normal regeneration and repair. Without these cells, the aniridic cornea does not heal well after surgery, and can become scarred and opaque even when no surgery has been done. This scarring is called "aniridic keratopathy," or sometimes, "corneal pannus" (for more information on these, see: [Aniridic keratopathy](#)) Because of this poor healing ability of the cornea, the use of contact lenses of any kind is now discouraged except in certain cases.

What is the difference between an Optometrist and an Ophthalmologist?

An optometrist is not a medical doctor, but is a doctor of optometry (O.D.). Optometrists diagnose vision problems and eye disease, prescribe eyeglasses and contact lenses, and prescribe drugs to treat eye disorders. They cannot perform surgery, but they often provide patients with pre- and postsurgical care. Sometimes ophthalmologists and optometrists work in the same practice and co-manage patients.

An ophthalmologist is a physician who specializes in the medical and surgical care of the eyes and visual system and in the prevention of eye disease and injury. They provide a full spectrum of care including routine eye exams, diagnosis and medical treatment of eye disorders and diseases, prescriptions for eyeglasses, surgery, and management of eye problems that are caused by systemic illnesses.

Eyecare Basics

Infants with aniridia should be referred for care by an ophthalmologist as soon as possible after diagnosis.

Eyecare for the infant or child with aniridia should include:

- 1.) Examinations approximately every 3 to 6 months --more often if complications such as glaucoma are identified.
- 2.) Thorough examination of all the structures of the eye. This examination may require EUA (Exam Under Anesthesia) during infancy and at periodic intervals throughout early childhood.
- 3.) Ocular photographs at the initial visit or EUA, if possible. Repeat if ocular changes occur.
- 4.) Tests for refraction (near or farsightedness) intraocular pressures (see Aniridia and Glaucoma) eye alignment, muscle strength, and other tests as necessary.

Your child's ophthalmologist should discuss with you: the importance of protecting your child's eyes from sunlight, signs and symptoms of infection, glaucoma, and whom to call if you have questions or suspect a problem.

Sometimes infants and children with aniridia require eyedrops for infections or glaucoma.

Because of the sensitivity and decreased healing ability of the aniridic cornea, you may want to remind your eye doctor that all eyedrops should be preservative-free whenever possible.

There are several ways to instill eyedrops with a minimum of fuss. Here are some suggestions from other parents:

- 1) Infants – wrap in a towel or blanket (to keep arms/hands down) Place child on his back on the floor, facing away from you, with his head between your knees. This will allow you to keep him from turning his head, while still having both hands free.
- 2) Place a drop in the corner of (closed) eye. Wait...when child opens the eye, the drop will go in.
- 3) Another method: instead of trying to push the eyelid up, try pulling the lower eyelid down. Place the drop inside the “pouch” created in the lowered lid, and release.

Aniridia And....

Nystagmus

Nystagmus is an involuntary eye movement. The degree and direction of eye movement can vary greatly from person to person.

U.S.A.: <http://www.nystagmus.org/>

Europe: <http://www.nystagmusnet.org/Links.htm>

Ptosis

Ptosis is an abnormally low position (drooping) of the upper eyelid. Ptosis occurs when the muscle that usually raises the eyelid is not strong enough to do so.

Ptosis is sometimes associated with aniridia. It may be present at birth, or noted within the first few months of life. Ptosis may also occur as a side effect of Vincristine, a drug commonly used to treat Wilms tumor.

Treatment of ptosis is usually surgery.

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ASOCIACION ESPANOLA DE –ANIRIDIA

Web: <http://www.aniridia.com>

Gêniris - formerly the French Aniridia Association

Web: <http://www.associationgeniris.free.fr/>

E-mail: aniridie.france@laposte.net

Aniridia Italiana

Web: <http://www.aniridia.it/>

E-mail: cqart@libero.it

Aniridia Liechtenstein

E-mail: maxped@adon.li

Asociación Mexicana de Aniridia

Web: <http://www.amda-ac.org/>

E-mail: yoannarcos@amda-ac.org

ANIRIDIA NORGE

Web: <http://www.aniridi.no/>

E-mail: khhansen@tiscali.no

Gillespie Syndrome

Web: <http://www.emedicine.com/OPH/topic317.htm>

Web: <http://www.gskasper.com/>

Email: kasper.gs@telia.com

American Foundation for the Blind
Provides information about local programs, including state Libraries for the Blind (Talking Books programs are free, call for information) and independent living skills programs.

11 Penn Plaza Suite 300
New York, NY 10001
PH: 800-232-5463
Web: <http://www.afb.org>

The Glaucoma Foundation
116 John Street, Suite 1605
New York, NY 10038
PH: 212-285-0080
Web: <http://www.glaucoma-foundation.org>

The New York Glaucoma Research Institute
Promotes research into causes of Glaucoma, development of alternative approaches to the treatment of Glaucoma, and development of a comprehensive patient information center about Glaucoma on the Internet.

The New York Eye and Ear Infirmary
310 East 14th Street
New York, NY 10003
Tel/Fax: 212-388-9517
Email: ritchmd@earthlink.net
Web: <http://www.glaucoma.net>

National Association for Parents of the Visually Impaired
Excellent source of information about parenting young children with VI.

P.O. Box 317
Watertown, MA 02247
PH: 617-972-7441
Web: <http://www.spedex.com/napvi>

National Institutes of Health: National Eye Institute
Information about current studies and trials of new or experimental
treatments for Aniridia or related problems, such as cataracts or glaucoma.

Building 31, Rm 6A32
31 Center Dr. MSC 2510
Bethesda, MD 20892-2510
PH: 301-496-5248
Web: <http://www.nei.nih.gov/>

"Children With Visual Impairments-A Parent's Guide"

Edited by M. Cay Holbrook, PhD.

Available through bookstores, or can be ordered through Exceptional
Parent Magazine (800-535-1910, order # WB1790D)

"Can't Your Child See?"

E. Scott.

Excellent guide to ideas for stimulating your visually impaired child's
development.

Available through bookstores, or can be ordered through Exceptional
Parent Magazine (800-535-1910, order #PE0410D)

Sunglasses for Children

Baby Banz - international website

Nonprescription sunglasses for children birth to 3yrs.

Order Online:

Web: <http://www.babybanz.com.au>

SoloBambini

Prescription and nonprescription glasses and sunglasses for children
birth to 7 yrs.

P.O. Box 349 Burlingame, California 94011

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