

Aniridia

What is Aniridia?

Aniridia means an absence of the iris or the colored part of the eye. A small rim of iris may be visible only with use of a special instrument by an ophthalmologist. The pupil is large and the condition is present in both eyes [See figure 1].

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. It is due to a defect in chromosome 11p. The PAX6 gene causes aniridia.

What is the chance of having another child with aniridia?

The chance of having a child with aniridia varies with the type of inheritance pattern responsible for the occurrence. With autosomal dominant transmission, there is a 50% chance of a child having aniridia with each pregnancy. When aniridia occurs for the first time in a family without affected parents (sporadic type of aniridia) the involved person has a 50% chance of having a child with aniridia with each pregnancy. A rare method of inheritance of aniridia is autosomal recessive in which a couple has a 25% chance of having a child with aniridia with each pregnancy. This type may be associated with mental retardation.

How does Aniridia affect vision?

Some children with aniridia have very good vision (20/30 or better) and some have poorer vision (worse than 20/200). All have absence of a pupil which causes excessive glare. Vision can be affected by a number of factors which have variable occurrence with aniridia. Some have scarring of the cornea (front of the eye) which causes light scatter. Cataracts can affect vision. The lens in the eye may be shifted out of its normal place (subluxated) and affect vision. Poor development of the fovea (responsible for fine vision) or the optic nerve can cause nystagmus (wobbly eyes) and possibly decrease vision. In addition 50% of people with aniridia develop glaucoma.

Is Aniridia associated with other health problems?

Aniridia may be present with no other systems of the body being affected or as a syndrome (a group of signs which occur consistently together). Aniridia can be associated with a kidney tumor called a Wilm's tumor (nephroblastoma). WAGR syndrome stands for Wilm's tumor, Aniridia, Genital abnormalities and Retardation (mental). Gillespie's syndrome is a combination of aniridia, mental retardation and balance problems (ataxia).



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

What treatment is available for aniridia?

Children with aniridia need regular eye exams to assess vision and need for glasses, for glaucoma, and for cataract. If glaucoma develops treatment is typically with eye drops. If vision is reduced by a cataract, removal of lens with or without intraocular lens implant may be indicated. A recent development is an artificial iris that is sometimes placed after cataract removal. Genetic counseling and examination of family members for subtle changes in the iris can sometimes help establish the mode of inheritance. When aniridia is first diagnosed it may be necessary to test for a kidney tumor. Genetic tests of the chromosomes can sometimes help determine the likelihood of a kidney tumor.

What type of glasses should a child with aniridia wear?

The eye doctor checks and prescribes glasses if indicated. Glasses to help cope with the glare and to protect the eye from sunlight and injury (polycarbonate lens) are often prescribed. Magnifying devices (low vision aids) may be helpful with reading.

What about contact lens use in aniridia?

Special painted soft contact lens are available that can reduce glare, improve cosmesis, and potentially improve vision. If surface of the eye (cornea) problems are present, the contact lens may not be indicated.

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 eye doctor.

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.

Are there any aniridia support groups?

Aniridia Foundation International <http://www.aniridia.net/>