



Aniridia At-A-Glance

Definition of Aniridia:

- "Aniridia" means "without iris."
- First indication is the absence of the colored part of the eye (iris).
- May appear as an enlarged pupil.

Characteristics of Aniridia:

- Defined by underdevelopment of iris tissue.
- Increased light sensitivity (photophobia) and glare issues.
- Pan-ocular condition affects other eye components, including the cornea, fovea, retina, optic nerve, anterior chamber, and lens.

Associated Ocular Conditions:

- Individuals may have one or more of the following conditions impacting vision: glaucoma, foveal hypoplasia, nystagmus, strabismus, dry eye, corneal degeneration, and cataracts.
- Some conditions related to aniridia are non-degenerative (do not get worse over time).
- Degenerative conditions include corneal keratopathy, aniridic glaucoma, and cataracts.

Vision and Variability:

- Degree of vision problems varies.
- Visual acuity for many is between 20/80 and 20/200 – but can vary widely within and outside this span.
- Some with aniridia are legally blind, while others can drive.
- Some with aniridia use Braille; others do not.

Other Medical Problems Associated:

- The following may be associated: glucose intolerance, central auditory processing disorder, decreased or absent sense of smell, and subtle brain structure abnormalities.
- Those with WAGR syndrome may have additional medical problems, including Wilms Tumor.

Diagnosis:

- Diagnosed by a pediatric ophthalmologist.
- Lack of pupil reaction to light observed during routine exams.
- Diagnosis confirmed through genetic testing by a geneticist.
- Genetic counselors associated with universities and children's hospitals can provide more information.

Prevalence:

- Affects approximately 1 in 60,000 births. Ocular problems associated with aniridia are common.