

ANIRIDIA

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ANIRIDIA

Common Associated Conditions

Condition and Description

Aniridia is a congenial condition that caused the eye to be undeveloped. A gene responsible for eye development does not function correctly and interferes with the development of the iris in the early months of fetal life. The baby is born with eye problems that vary among individuals affected with this condition. Typically they will have very little iris (iris hypoplasia), and the tissue that is there will form a collar around the pupil of the eye, thus not constricting in bright light. There are many variations of aniridia that range from mild to severe. Vision may be limited or greatly reduced, depending on what other conditions affect the eyes.

Parents will often detect this condition when the baby is very sensitive to light and the pupil of the eye seems very large.

Prevalence

This is a very rare disorder, occurring in 1 of 60,000-90,000 live births. Aniridia is inherited as a dominant gene, and therefore each child born to a parent with the condition will have a 50% chance of having the gene. The gene expression may vary, however, so that each family member may have a somewhat different type of disorder. It is bilateral, meaning that both eyes will be affected. In 85% of cases there will be a family history, but 15% of the time there will be a sporadic genetic change that causes the condition.

The most common associated condition is involuntary movement of the eyes (nystagmus), occurring in nearly 90% of children. They may also have cloudy lens of the eye (cataract), occurring in 85% of children, as well as increased pressure in the eye (glaucoma), in 70%). Other conditions include cloudy cornea (aniridic keratopathy), underdevelopment of the retina at the back of the eye (fovea or macular hypoplasia), underdevelopment of the optic nerve (optic nerve hypoplasia), loss of vision of the weaker eye (amblyopia), small cornea (microcornea), and crossing or wandering of the eye (strabismus). Children with aniridia may have an impaired sense of smell and also an intellectual disorder called dysnomia (difficulty recalling words even though they understand the meaning of the words).

Aniridia is a part of two very rare genetic syndromes: WAGR and Gillespie syndrome. The same gene is responsible for Wilm's tumor so children with aniridia must be carefully screened for this serious tumor of the adrenal gland on the kidney that can develop in early childhood. Thirty percent of children develop Wilm's tumor before age 5 years, so it is very important that they be carefully followed.

Short-term Treatment and Outcomes

Genetic counseling will be offered to the family of a child with aniridia. Short-term, children with aniridia can do very well, though they need to be carefully followed by an ophthalmologist throughout life so that any complications are diagnosed and promptly



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treated. They will typically have about 20/100 vision, though this varies greatly between individuals. They can develop esotropia (turning in of the eye), and if this is not promptly treated, amblyopia occurs where vision is rapidly lost in the weaker eye. With careful treatment children with aniridia will have a very good outcome although there is no cure and they will need to have assistance for adapting to living with reduced vision.

Long-term Treatment and Outcomes

Vision typically gets worse with age as cataracts progress (though these can be treated with surgery and contact lenses. Glaucoma frequently develops in individuals with aniridia, affecting the function of optic nerve over time and further reducing vision. Ongoing screening for Wilm's tumor is essential.

Individuals with aniridia will greatly benefit from being exposed to others who have successfully lived full lives with limited vision. There are a great many adaptive devices and techniques that can assist individuals to cope with low vision or blindness. Information and support groups are available on the internet.

Common Complications

Corneal opacification (clouding) also occurs over time, and some people benefit from corneal transplantation. Retinal problems can progress, as can the optic nerve damage.

Implications for Children's Development

Most children with aniridia will have normal intelligence, except for the very few who have complex syndromes. The main developmental effect will be to learn to be self-sufficient and fully participatory in life activities with low vision that might gradually progress to blindness. Families where multiple members are affected with this disorder will cope with this condition quite differently from those where it occurs sporadically with no family history. Even in families where others are affected, the parents will always hope that this child will be born with normal eyes. A period of grieving is expected, and the family will require support and information. Treatment in a tertiary center that has extensive experience with aniridia is desirable, as new techniques are developed constantly to treat eye disorders and to cope with low vision.

Building children's self-esteem and independence will be essential so that they will learn to incorporate this possibly progressive disorder into a normal life style. Many resources are available online and in the community to support the parents and children with aniridia.

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