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My46 Trait Profile

Aniridia

Aniridia is the congenital (present at birth) absence of the colored part of the eye, called the iris. It is caused by changes in the PAX6 gene. Aniridia may occur as an isolated finding, or as part of a syndrome with additional medical issues.

Characteristics of Aniridia

Aniridia is the congenital (present at birth) absence of the colored part of the eye, called the iris. The iris may be missing completely or it may be much smaller than usual. In most cases, both eyes are affected. This iris is responsible for controlling the amount of light that enters the eye. People with aniridia are not able to control this very well, so they may be more sensitive to lights than people without aniridia.

People with aniridia may have additional problems with their eyes. Glaucoma (increased pressure in the eye) or cataracts (cloudiness in the lens of the eye) might develop. The eyes might bounce or shake back and forth; this movement is called nystagmus. Light sensing cells in the back of the eye might be under-developed (foveal hypoplasia), and thus, people with aniridia may have less clear central vision. This may make it difficult to see certain details in the environment. Affected individuals may be near-sighted or far-sighted, or their eyes may not align properly. Vision may get worse over time.

Diagnosis/Testing

Most people with aniridia have a change or mutation in a gene called PAX6. The PAX6 gene makes a protein called a transcription factor. Transcription factors attach to DNA to control how other genes are used in the body. The PAX6 gene plays an early role in the development of human eyes, parts of the brain and spinal cord, and pancreas.

Management/Surveillance

People with aniridia should be followed by an ophthalmologist (medical eye doctor) to monitor for eye complications. Glasses may be necessary to correct vision. Due to photosensitivity (sensitivity to light), affected individuals may benefit from tinted contact lenses or sunglasses. People with aniridia may need low-vision aids or vision therapy to adapt to their impaired sense of sight.

Mode of inheritance

Aniridia caused by PAX6 gene mutations is inherited in an autosomal dominant pattern. This means having one PAX6 gene mutation is enough for an individual to be affected. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not the individual with aniridia has a parent affected with aniridia. If a parent also has aniridia due to a PAX6 gene mutation, the risk of having a child with aniridia is 50% with each pregnancy. If a parent does not have aniridia, the risk of other siblings being affected is very low.

Special considerations

Some people with aniridia have additional health problems that affect other parts of the body. Because certain symptoms are known to cluster together, the group of symptoms is called a syndrome.

WAGR syndrome is named for its common features: Wilms tumor (childhood kidney cancer), Aniridia, Genitourinary abnormalities, and mental Retardation (more commonly referred to as intellectual disability). It is important for the care team to distinguish between isolated aniridia and WAGR syndrome due to the risk for cancer. Genetic testing can help make the correct diagnosis, especially in very young children who might only have aniridia at the time of evaluation. (See separate trait profile for “WT1-related Wilms tumor” for more information).

Resources

The Vision for Tomorrow Foundation
http://www.visionfortomorrow.org/aniridia/
Genetics Home Reference: Aniridia
http://ghr.nlm.nih.gov/condition/aniridia

References

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