Fundus Albipunctatus

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Fundus albipunctatus is an eye disorder characterized by an impaired ability to see in low light (night blindness) and the presence of whitish-yellow flecks in the retina, which is the specialized light-sensitive tissue in the inner lining of the back of the eye (the fundus). The flecks are detected during an eye examination.

Keywords: genetic conditions

1. Introduction

Individuals with fundus albipunctatus experience night blindness from an early age. In particular, they have delayed dark adaptation, which means they have trouble adapting from bright light to dark conditions, such as when driving into a dark tunnel on a sunny day. It often takes hours for adaptation to occur. Their vision in bright light is usually normal.

The flecks are especially abundant near the outer edge (the periphery) of the retina. Their density varies among affected individuals; some people have numerous flecks that overlap, while others have fewer. For unknown reasons, the flecks get smaller or fade with age in some affected individuals, although night vision does not improve.

While fundus albipunctatus typically does not worsen (progress) over time, some individuals with the condition develop other eye conditions, such as breakdown of the central region of the retina known as the macula (macular degeneration) with loss of specialized light receptor cells called cones, which can affect vision in bright light.

2. Frequency

Fundus albipunctatus is a rare disorder. Its prevalence is unknown.

3. Causes

Fundus albipunctatus is primarily caused by mutations in the *RDH5* gene. This gene is involved in a multi-step process called the visual cycle, by which light entering the eye is converted into electrical signals that are interpreted as vision. In rare cases, fundus albipunctatus is caused by mutations in other genes that play roles in the visual cycle.

An integral operation of the visual cycle is the recycling of a molecule called 11-cis retinal, which is a form of vitamin A that is needed for the conversion of light to electrical signals. The *RDH5* gene provides instructions for making an enzyme called 11-cis retinol dehydrogenase 5, which performs one step in this recycling process. This enzyme converts a molecule called 11-cis retinol to 11-cis retinal.

RDH5 gene mutations are thought to reduce or eliminate the function of the 11-cis retinol dehydrogenase 5 enzyme, which results in a shortage of 11-cis retinal. Without this important molecule, electrical signals integral for vision are not stimulated, and vision is impaired.

For vision in low-light conditions, the eyes primarily use 11-cis retinol dehydrogenase 5 to generate 11-cis retinal, which is why a shortage of this enzyme's function impairs night vision.

Researchers speculate that impairment of 11-cis retinol dehydrogenase 5 also leads to the accumulation of 11-cis retinol and related molecules, forming the flecks characteristic of fundus albipunctatus.

3.1. The genes associated with Fundus albipunctatus

- RDH5
- RPE65

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- albipunctate retinal dystrophy
- Lauber's disease
- pigmentary retinal dystrophy

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