RDH5 Gene

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retinol dehydrogenase 5

genes

1. Normal Function

The *RDH5* gene provides instructions for making an enzyme called 11-cis retinol dehydrogenase 5, which is necessary for normal vision, especially in low-light conditions (night vision). This enzyme is found in a thin layer of cells at the back of the eye called the retinal pigment epithelium (RPE). This cell layer supports and nourishes the retina, which is the light-sensitive tissue in the inner lining of the back of the eye (the fundus).

11-cis retinol dehydrogenase 5 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. 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 retinol dehydrogenase 5 enzyme converts a molecule called 11-cis retinol to 11-cis retinal. In light-sensing cells in the retina known as photoreceptors, 11-cis retinal combines with a protein called an opsin to form a photosensitive pigment. When light hits this pigment, 11-cis retinal is altered, forming another molecule called all-trans retinal. This conversion triggers a series of chemical reactions that create electrical signals. 11-cis retinol dehydrogenase 5 then helps convert all-trans retinal back to 11-cis retinal so the visual cycle can begin again.

The eyes contain two types of photoreceptors, rods and cones. Rods are needed for vision in low light, while cones are needed for vision in bright light, including color vision. Rods primarily use 11-cis retinol dehydrogenase 5 to generate 11-cis retinal. Cones also use 11-cis retinol dehydrogenase 5, but they are thought to have additional pathways to produce 11-cis retinal.

2. Health Conditions Related to Genetic Changes

2.1. Fundus albipunctatus

At least 48 mutations in the *RDH5* gene have been found to cause fundus albipunctatus, a condition characterized by impaired night vision and whitish-yellow flecks in the retina. The *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 in photoreceptors, electrical signals integral for vision are not stimulated, and vision is impaired. Because rods rely on 11-cis retinol dehydrogenase 5 for 11-cis retinal production, vision in low light is particularly affected by impairment of this enzyme's function. 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. Other Names for This Gene

- 11-cis RDH
- 11-cis retinol dehydrogenase precursor
- 11-cis RoDH
- 9-cis retinol dehydrogenase
- 9-cis-retinol specific dehydrogenase
- 9cRDH
- HSD17B9
- RDH1
- retinol dehydrogenase 1
- retinol dehydrogenase 5 (11-cis and 9-cis)
- retinol dehydrogenase 5 (11-cis/9-cis)
- SDR9C5
- short chain dehydrogenase/reductase family 9C member 5

References

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