ABCA4 Gene

Subjects: Genetics & Heredity

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ATP binding cassette subfamily A member 4

genes

1. Normal Function

The *ABCA4* gene provides instructions for making a protein that is found in the retina, the specialized light-sensitive tissue that lines the back of the eye. Specifically, the ABCA4 protein is produced in the retina's light receptor cells (photoreceptors). The ABCA4 protein is active following phototransduction, the process by which light entering the eye is converted into electrical signals that are transmitted to the brain. Phototransduction leads to the formation of potentially toxic substances that can damage photoreceptor cells. The ABCA4 protein removes one of these substances, called N-retinylidene-PE, from photoreceptor cells.

2. Health Conditions Related to Genetic Changes

2.1 Cone-rod dystrophy

More than 40 mutations in the *ABCA4* gene have been found to cause a vision disorder called cone-rod dystrophy. The problems associated with this condition include a loss of visual sharpness (acuity), an increased sensitivity to light (photophobia), and impaired color vision. These vision problems worsen over time. It is estimated that *ABCA4* gene mutations account for 30 to 60 percent of cases of cone-rod dystrophy that are inherited in an autosomal recessive pattern, which means that both copies of the gene in each cell have mutations.

Most of the *ABCA4* gene mutations that cause cone-rod dystrophy change single protein building blocks (amino acids) in the ABCA4 protein. The altered protein cannot remove N-retinylidene-PE from photoreceptor cells. As a result, N-retinylidene-PE combines with another substance to produce a molecule called N-retinylidene-N-retinylethanolamine (A2E), which builds up in these cells. The buildup of A2E is toxic to photoreceptor cells and leads to their deterioration, causing progressive vision loss in people with cone-rod dystrophy. Cone-rod dystrophy caused by *ABCA4* gene mutations tends to be associated with more severe vision problems than cone-rod dystrophy caused by other genetic mutations.

2.2 Stargardt macular degeneration

More than 640 mutations in the *ABCA4* gene have been found to cause Stargardt macular degeneration. Mutations in this gene are the most common cause of this eye disease. Stargardt macular degeneration is characterized by vision loss that worsens over time, particularly affecting central and night vision. Most of these mutations change amino acids in the ABCA4 protein. A malfunctioning ABCA4 protein cannot remove N-retinylidene-PE from photoreceptor cells. As a result, N-retinylidene-PE combines with another substance to produce a fatty yellow pigment called lipofuscin, which builds up in retinal cells. The buildup of lipofuscin is toxic to the cells of the retina and causes progressive vision loss in people with Stargardt macular degeneration.

Several *ABCA4* gene mutations have been found to cause different forms of vision loss in different individuals. It is unclear how mutations in the *ABCA4* gene can cause different eye disorders.

2.3 Age-related macular degeneration

MedlinePlus Genetics provides information about Age-related macular degeneration

2.4 Retinitis pigmentosa

MedlinePlus Genetics provides information about Retinitis pigmentosa

3. Other Names for This Gene

- ABCA4_HUMAN
- ABCR
- ATP-binding cassette sub-family A member 4
- ATP-binding cassette transporter, retinal-specific
- ATP-binding cassette, sub-family A (ABC1), member 4
- photoreceptor rim protein
- retina-specific ABC transporter
- retinal-specific ATP-binding cassette transporter
- RIM ABC transporter
- RIM protein
- RMP

References

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