# ABCA4 gene

ATP binding cassette subfamily A member 4

## 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-sensing cells (photoreceptors). The ABCA4 protein transports potentially toxic substances that can damage photoreceptors. These substances form after phototransduction, the process by which light entering the eye is converted into electrical signals that are transmitted to the brain. The ABCA4 protein removes one of these substances, called N-retinylidene-PE, from photoreceptors.

## Health Conditions Related to Genetic Changes

Cone-rod dystrophy

Many variants (also called 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 variants account for 30 to 60 percent of cases of cone-rod dystrophy that are inherited in an autosomal recessive pattern, which means both copies of the gene in each cell must have a variant to cause the disorder.

Most of the *ABCA4* gene variants 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 photoreceptors. 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 photoreceptors and leads to their deterioration, causing progressive vision loss in people with cone-rod dystrophy. Cone-rod dystrophy caused by *ABCA4* gene variants tends to be associated with more severe vision problems than cone-rod dystrophy caused by other genetic variants.

Stargardt macular degeneration

More than 1,000 variants in the *ABCA4* gene have been found to cause Stargardt

macular degeneration. Variants in this gene are the most common cause of this eye disease. Stargardt macular degeneration is characterized by vision loss that worsens over time.

Most of these variants change amino acids in the ABCA4 protein. A malfunctioning ABCA4 protein cannot remove N-retinylidene-PE from photoreceptors. 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.

Age-related macular degeneration

MedlinePlus Genetics provides information about Age-related macular degeneration

Retinitis pigmentosa

MedlinePlus Genetics provides information about Retinitis pigmentosa

## 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

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ABCA4 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=24[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ABCA4%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ATP-BINDING CASSETTE, SUBFAMILY A, MEMBER 4; ABCA4 (https://omim.org/entry/601691)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/24)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ABCA4[gene])

## References


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## Genomic Location

The *ABCA4* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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