CNGB3 Gene

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cyclic nucleotide gated channel beta 3

genes

1. Normal Function

The *CNGB3* gene provides instructions for making one part (the beta subunit) of the cone photoreceptor cyclic nucleotide-gated (CNG) channel. These channels are found exclusively in light-detecting (photoreceptor) cells called cones, which are located in a specialized tissue at the back of the eye known as the retina. Cones provide vision in bright light (daylight vision), including color vision. Other photoreceptor cells, called rods, provide vision in low light (night vision).

CNG channels are openings in the cell membrane that transport positively charged atoms (cations) into cells. In cones, CNG channels remain open under dark conditions, allowing cations to flow in. When light enters the eye, it triggers the closure of these channels, stopping the inward flow of cations. This change in cation transport alters the cone's electrical charge, which ultimately generates a signal that is interpreted by the brain as vision. This process of translating light into an electrical signal is called phototransduction.

2. Health Conditions Related to Genetic Changes

2.1. Achromatopsia

More than 40 mutations in the *CNGB3* gene have been found to cause the vision disorder achromatopsia. These mutations cause 50 to 70 percent of cases of complete achromatopsia, a form of the disorder characterized by a total lack of color vision and other vision problems that are present from early infancy. Worldwide, the most common mutation that causes this condition deletes a single DNA building block (base pair) from the *CNGB3* gene. This mutation can be written as 1148delC.

Complete achromatopsia occurs frequently in Pingelapese islanders, who live on one of the Eastern Caroline Islands of Micronesia. Among the Pingelapese, this condition results from a mutation that changes a single protein building block (amino acid) in the beta subunit. This mutation replaces the amino acid serine with the amino acid phenylalanine at position 435 in the protein (written as Ser435Phe or S435F).

Most *CNGB3* gene mutations prevent the production of any functional beta subunit, which alters the structure of CNG channels. The resulting channels are nonfunctional and prevent cones from carrying out phototransduction. Researchers speculate that the defective channels allow a huge influx of cations into cones, which ultimately causes these cells to self-destruct (undergo apoptosis). A loss of cone function underlies the lack of color vision and other vision problems in people with complete achromatopsia.

Because these CNG channels are specific to cones, rods are generally unaffected by this disorder.

2.2. Cone-Rod Dystrophy

Cone-rod dystrophy

2.3. Other Disorders

Mutations in the *CNGB3* gene have also been identified in a small percentage of cases of progressive cone dystrophy. Like achromatopsia (described above), this condition affects the function of cones in the retina. However, unlike achromatopsia, progressive cone dystrophy is associated with cones that work normally at birth but begin to malfunction in childhood or adolescence. Over time, people with progressive cone dystrophy develop increasing blurriness and loss of color vision. It is unclear why some *CNGB3* gene mutations cause achromatopsia and others result in progressive cone dystrophy.

3. Other Names for This Gene

- ACHM3
- CNGB3_HUMAN
- cone photoreceptor cGMP-gated cation channel beta-subunit
- cyclic nucleotide-gated cation channel modulatory subunit

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