GNAT2 Gene

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Contributor: Dean Liu

G protein subunit alpha transducin 2

genes

1. Introduction

The *GNAT2* gene provides instructions for making one part (called the cone-specific alpha subunit) of a protein called transducin. This protein is found 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).

Transducin plays an essential role in transmitting visual signals from photoreceptor cells in the retina to the brain through a process called phototransduction. Photoreceptors contain special pigments (called photopigments) that absorb light. The photopigments activate transducin, which triggers a series of chemical reactions within the cell. These reactions alter the cell's electrical charge, ultimately generating a signal that is interpreted by the brain as vision.

2. Health Conditions Related to Genetic Changes

2.1. Achromatopsia

At least 10 mutations in the *GNAT2* gene have been found to cause the vision disorder achromatopsia. These mutations are a relatively uncommon cause 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. *GNAT2* gene mutations have also been identified in a few individuals with incomplete achromatopsia, a milder form of the disorder associated with limited color vision.

The *GNAT2* gene mutations that underlie complete achromatopsia lead to an abnormally small, nonfunctional version of the cone-specific alpha subunit of transducin. Without this subunit, cones have no functional transducin, and

they are unable to carry out phototransduction. (The subunit produced from the GNAT2 gene is specific to cones, so rods are typically unaffected by this disorder.) A loss of cone function underlies the lack of color vision and other

vision problems in people with complete achromatopsia.

At least one known *GNAT2* gene mutation causes incomplete achromatopsia. The mutation, which is written as c.461+2G>A, affects the way the gene's instructions are pieced together to form the subunit protein. This mutation allows the production of some functional cone-specific alpha subunit, although the amount of the subunit is greatly reduced. As a result, a small amount of functional transducin is available to play its role in phototransduction, and the partially functioning cones can transmit some visual information to the brain.

3. Other Names for This Gene

- ACHM4
- cone-type transducin alpha subunit
- GNAT2_HUMAN
- GNATC
- guanine nucleotide binding protein (G protein), alpha transducing activity polypeptide 2
- guanine nucleotide binding protein, alpha transducing activity polypeptide 2
- transducin alpha-2 chain
- transducin, cone-specific, alpha polypeptide

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