GNAT1 gene

G protein subunit alpha transducin 1

Normal Function

The *GNAT1* gene provides instructions for making a protein called alpha (α)-transducin. This protein is one part (the alpha subunit) of a protein complex called transducin. There are several versions of transducin made up of different subunits. Each version is found in a particular cell type in the light-sensitive tissue at the back of the eye (the retina), where it plays a role in transmitting visual signals from the eye to the brain.

The transducin complex that contains α-transducin is found only in specialized light receptor cells in the retina called rods. Rods are responsible for vision in low-light conditions. When light enters the eye, a rod cell protein called rhodopsin is turned on (activated), which then activates α-transducin. Once activated, α-transducin breaks away from the transducin complex in order to activate another protein called cGMP-PDE, which triggers a series of chemical reactions that create electrical signals. These signals are transmitted from rod cells to the brain, where they are interpreted as vision.

Health Conditions Related to Genetic Changes

Autosomal dominant congenital stationary night blindness

At least two mutations in the *GNAT1* gene have been found to cause autosomal dominant congenital stationary night blindness, which is characterized by the inability to see in low light.

One of these mutations impairs the protein's ability to activate cGMP-PDE; the other mutation results in a protein that is constantly turned on (constitutively activated). Both of these mutations disrupt the pathway that creates visual signals to be sent from rod cells to the brain. A nonfunctional α-transducin protein stops the signaling pathway. When α-transducin is constitutively activated, the signals that the rod cells send to the brain are constantly occurring, even in bright light. Visual information from rod cells is then perceived by the brain as not meaningful, resulting in night blindness.

Other Names for This Gene

- CSNBAD3
- GBT1

- GNAT1_HUMAN
- GNATR
- guanine nucleotide binding protein (G protein), alpha transducing activity polypeptide 1
- guanine nucleotide-binding protein G(t) subunit alpha-1
- guanine nucleotide-binding protein G(T), alpha-1 subunit
- rod-type transducin alpha subunit
- transducin alpha-1 chain
- transducin, rod-specific

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GNAT1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2779[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28GNAT1%5BTIAB%5D%29+OR+%28%28transducin+alpha+subunit%5BTIAB%5D%29+OR+%28transducin+alpha-1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GUANINE NUCLEOTIDE-BINDING PROTEIN, ALPHA-TRANSDUCING ACTIVITY POLYPEPTIDE 1; GNAT1 (https://omim.org/entry/139330)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2779)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GNAT1[gene])

References

- Dryja TP, Hahn LB, Reboul T, Arnaud B. Missense mutation in the gene encodingthe alpha subunit of rod transducin in the Nougaret form of congenital stationarynight blindness. Nat Genet. 1996 Jul;13(3):358-60. doi: 10.1038/ng0796-358. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/8673138)
- Sandberg MA, Pawlyk BS, Dan J, Arnaud B, Dryja TP, Berson EL. Rod and

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- Szabo V, Kreienkamp HJ, Rosenberg T, Gal A. p.Gln200Glu, a putativeconstitutively active mutant of rod alpha-transducin (GNAT1) in autosomaldominant congenital stationary night blindness. Hum Mutat. 2007 Jul;28(7):741-2.doi: 10.1002/humu.9499. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17584859)

Genomic Location

The *GNAT1* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

Last updated November 1, 2013