CHRNB2 gene

cholinergic receptor nicotinic beta 2 subunit

Normal Function

The *CHRNB2* gene provides instructions for making one part (subunit) of a larger protein called a neuronal nicotinic acetylcholine receptor (nAChR). Each nAChR protein is made up of a combination of five subunits, usually two alpha (α) and three beta (β) subunits. Many different combinations are possible, and the characteristics of each nAChR protein depend on which subunits it contains. In the brain, nAChR proteins most commonly consist of two α4 subunits and three β2 subunits. The *CHRNB2* gene is responsible for producing the β2 subunit.

In the brain, nAChR proteins are widely distributed and play an important role in chemical signaling between nerve cells (neurons). The nAChR proteins act as channels, allowing charged atoms (ions) including calcium, sodium, and potassium to cross the cell membrane. These channels open when attached to a brain chemical (neurotransmitter) called acetylcholine. The channels also open in response to nicotine, the addictive substance in tobacco.

Communication between neurons depends on neurotransmitters, which are released from one neuron and taken up by neighboring neurons. The release and uptake of these chemicals are tightly regulated to ensure that signals are passed efficiently and accurately between neurons. Researchers believe that nAChR channels play an important role in controlling the normal release and uptake of neurotransmitters.

A wide range of brain functions depend on nAChR channels, including sleep and arousal, fatigue, anxiety, attention, pain perception, and memory. The channels are also active before birth, which suggests that they are involved in early brain development. At least one drug that targets nAChR channels in the brain has been developed to help people quit smoking; other medications targeting these channels are under study for the treatment of schizophrenia, Alzheimer's disease, and pain.

Health Conditions Related to Genetic Changes

Autosomal dominant nocturnal frontal lobe epilepsy

At least three mutations in the *CHRNB2* gene have been identified in people with autosomal dominant nocturnal frontal lobe epilepsy (ADNFLE). Each of these mutations

changes a single protein building block (amino acid) in the β2 subunit of nAChR channels.

CHRNB2 mutations make nAChR channels more sensitive to the neurotransmitter acetylcholine, allowing the channels to open more easily than usual. The resulting increase in ion flow across the cell membrane alters the release of neurotransmitters, which changes signaling between neurons. Researchers believe that the overexcitement of certain neurons in the brain triggers the abnormal brain activity associated with seizures. It is unclear why the seizures seen in ADNFLE start in the frontal lobes of the brain and occur most often during sleep.

Other Names for This Gene

- Acetylcholine receptor, neuronal nicotinic, beta-2 subunit
- ACHB2_HUMAN
- cholinergic receptor, nicotinic beta 2
- cholinergic receptor, nicotinic, beta 2 (neuronal)
- cholinergic receptor, nicotinic, beta polypeptide 2 (neuronal)
- EFNL3
- nAChRB2
- neuronal nicotinic acetylcholine receptor beta 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CHRNB2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1141[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28CHRNB2%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, BETA POLYPEPTIDE 2; CHRNB2 (https://omim.org/entry/118507)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1141)

- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CHRNB2[gene])

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Genomic Location

The *CHRNB2* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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