GNAI3 gene

G protein subunit alpha i3

Normal Function

The *GNAI3* gene provides instructions for making one part, the *GNAI3* inhibitory alpha subunit, of a protein complex called a guanine nucleotide-binding protein (G protein). G proteins are composed of three protein subunits: alpha, beta, and gamma. Each of these subunits is produced from a different gene.

Through a process called signal transduction, G proteins trigger a complex network of signaling pathways within cells. These pathways help transmit information from the outside of the cell to the inside of the cell. Specifically, G proteins that are made with the *GNAI3* inhibitory alpha subunit reduce (inhibit) the activity of an enzyme called adenylyl cyclase. This enzyme is important for transmitting information within cells. By influencing many cell activities, G protein signaling helps cells grow and divide or take on specialized functions.

Studies suggest that G protein signaling that involves the *GNAI3* inhibitory alpha subunit contributes to the development of the first and second pharyngeal arches. During early development, these structures develop into the bones and tissues of the head and face, including the jawbones, facial muscles, and inner and outer ears.

Health Conditions Related to Genetic Changes

Auriculocondylar syndrome

Variants (also called mutations) in the *GNAI3* gene have been found to cause auriculocondylar syndrome. This disorder affects the development of various facial features, primarily the ears and lower jaw (mandible). Many of the *GNAI3* gene variants that cause auriculocondylar syndrome lead to the substitution of one protein building block (amino acid) for another in the *GNAI3* inhibitory alpha subunit. These variants likely alter the structure of the *GNAI3* inhibitory alpha subunit, impairing G protein signaling and disrupting the development of the structures that are formed from the first and second pharyngeal arches.

Other Names for This Gene

- 87U6

- ARCND1
- ARCODS
- guanine nucleotide binding protein (G protein), alpha inhibiting activity polypeptide 3

- guanine nucleotide binding protein, alpha-inhibiting activity polypeptide 3
- guanine nucleotide-binding protein G(k) subunit alpha
- HG1A

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GNAI3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2773[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28GNAI3%5BTIAB%5D%29+OR+%28%28guanine+nucleotide+binding+protein%5BTIAB%5D%29+AND+%28alpha+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28achromatopsia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- GUANINE NUCLEOTIDE-BINDING PROTEIN, ALPHA-INHIBITING ACTIVITY POLYPEPTIDE 3; GNAI3 (https://omim.org/entry/139370)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2773)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GNAI3[gene])

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Genomic Location

The *GNAI3* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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