

## GNAI3 gene

G protein subunit alpha i3

### Normal Function

The *GNAI3* gene provides instructions for making one component, the 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 outside the cell to inside the cell. Specifically, G proteins made with the *GNAI3* inhibitory alpha subunit reduce (inhibit) the activity of an enzyme called adenylyl cyclase, which is an important chemical messenger within cells. G protein signaling ultimately influences many cell activities, instructing the cell to grow, divide, or take on specialized functions.

Studies suggest that G protein signaling involving the *GNAI3* inhibitory alpha subunit contributes to the development of the first and second pharyngeal arches. These embryonic structures ultimately develop into the jawbones, facial muscles, middle ear bones, ear canals, outer ears, and related tissues.

### Health Conditions Related to Genetic Changes

#### Auriculo-condylar syndrome

At least two mutations in the *GNAI3* gene have been found to cause auriculo-condylar syndrome, a disorder that primarily affects the development of the ears and lower jaw (mandible). The identified mutations change single protein building blocks (amino acids) in the inhibitory alpha subunit. These mutations likely alter the structure of the inhibitory alpha subunit and impair G protein signaling. Abnormal signaling alters the formation of the lower jaw: instead of developing normally, the lower jaw becomes shaped more like the smaller upper jaw (maxilla). The abnormal shape leads to an unusually small chin (micrognathia) and problems with jaw function. Researchers are working to determine how mutations in this gene lead to the other developmental abnormalities associated with auriculo-condylar syndrome.

## Other Names for This Gene

- 87U6
- ARCND1
- g(i) alpha-3
- GNAI3\_HUMAN
- guanine nucleotide binding protein (G protein), alpha inhibiting activity polypeptide 3
- guanine nucleotide-binding protein G(k) subunit alpha

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of GNAI3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2773\[geneid\]](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%29>)

### 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\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GNAI3[gene]))

## References

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## Genomic Location

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

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