

Product datasheet for **RC402263**

G protein alpha S (GNAS) (NM_000516) Human Mutant ORF Clone

Product data:

| | |
|---------------------------|---|
| Product Type: | Mutant ORF Clones |
| Product Name: | G protein alpha S (GNAS) (NM_000516) Human Mutant ORF Clone |
| Mutation Description: | A102E |
| Affected Codon#: | 102 |
| Affected NT#: | 305 |
| Nucleotide Mutation: | GNAS Mutant (A102E), Myc-DDK-tagged ORF clone of Homo sapiens GNAS complex locus (GNAS), transcript variant 1 as transfection-ready DNA |
| Effect: | Albrih herediry oseodysrophy |
| Symbol: | GNAS |
| Synonyms: | AHO; C20orf45; GNAS1; GPSA; GSA; GSP; NESP; PITA3; POH; SCG6; SgVI |
| E. coli Selection: | Kanamycin (25 ug/mL) |
| Mammalian Cell Selection: | Neomycin |
| Vector: | pCMV6-Entry (PS100001) |
| Tag: | Myc-DDK |
| ACCN: | NM_000516 |
| ORF Size: | 1182 bp |
| Restriction Sites: | Sgfl-NotI |



[View online »](#)

ORF Nucleotide Sequence:

>RC402263 representing NM_000516
 Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC
 GCC**GCGATCGCC**

ATGGGCTGCCTCGGGAACAGTAAGACCGAGGACCAGCGCAACGAGGAGAAGGCGCAGCGTGAGGCCAACA
 AAAAGATCGAGAAGCAGCTGCAGAAGGACAAGCAGGTCTACCGGCCACGCACCGCCTGCTGCTGCTGGG
 TGCTGGAGAATCTGGTAAAAGCACCAATTGTGAAGCAGATGAGGATCCTGCATGTTAATGGTTTTAATGGA
 GAGGGCGGCGAAGAGGACCCGCAGGCTGCAAGGAGCAACAGCGATGGTGAGAAGGCAACCAAAAGTGCAGG
 ACATCAAAAACAACTGAAAGAGGAGATTGAAACCATTGTGGCCGCCATGAGCAACCTGGTGCCCCCGT
 GGAGCTGGCAACCCCGAGAACCAGTTCAGAGTGGACTACATCCTGAGTGTGATGAACGTGCCTGACTTT
 GACTTCCTCCCGAATTCATGAGCATGCCAAGGCTCTGTGGGAGGATGAAGGAGTGCCTGCCTGCTACG
 AACGCTCAACGAGTACCAGCTGATTGACTGTGCCAGTACTTCTGGACAAGATCGACGTGATCAAGCA
 GGCTGACTATGTGCCGAGCGATCAGGACCTGCTTCGCTGCCGTGCTCTGACTTCTGGAATCTTTGAGACC
 AAGTTCAGGTGGACAAAGTCAACTTCCACATGTTTGACGTGGGTGGCCAGCGCGATGAACGCCGCAAGT
 GGATCCAGTGTCTCAACGATGTGACTGCCATCATCTTCGTGGTGGCCAGCAGCAGCTACAACATGGTCAT
 CCGGGAGGACAACCAGACCAACCGCCTGCAGGAGGCTCTGAACCTCTCAAGAGCATCTGGAACAACAGA
 TGGCTGCGCACCATCTCTGTGATCCTGTTCCCAACAAGCAAGATCTGCTCGCTGAGAAAGTCTTTGCTG
 GGAAATCGAAGATTGAGGACTACTTCCAGAATTTGCTCGCTACACTACTCTGAGGATGCTACTCCCGA
 GCCCGGAGAGGACCCACGCGTGACCCGGGCCAAGTACTTCATTCGAGATGAGTTTCTGAGGATCAGCACT
 GCCAGTGGAGATGGGCGTCACTACTGCTACCCTCATTTACCTGCGCTGTGGACTGAGAACATCCGCC
 GTGTGTTCAACGACTGCCGTGACATCATTCAGCGCATGCACCTTCGTCACTACGAGCTGCTC

AG**GCGACCG**ACGCGTACGCGGCCGCTCGAGCAGAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCC
 TGGATTACAAGGATGACGACGA TAAGGTTTAA

Protein Sequence:

>RC402263 representing NM_000516
 Red=Cloning site Green=Tags(s)

MGCLGNSKTEDQRNEEKAQREANKKIEKQLQKDKQVYRATHRLLLLGAGESGKSTIVKQMRILHVNGFNG
 EGGEEDPQAARSNSDGEKATKVQDIKNNLKEE IETIVAAMSNLVPPVELANPENQFRVDYILSVMNVPDF
 DFPPEFYEHAKALWEDEGVRACYERSNEYQLIDCAQYFLDKIDVIKQADYVPSDQLLRVLTSGIFET
 KFQVDKVNFMFDVGGQRDERRKIQCNDVTAIIFVVASSSYNMVIREDNQTNRLQEALNLFKSIWNNR
 WLRTISVILFLNKQDLLAEKVLAGKSKIEDYFPEFARYTTPEDATPEPGEPRVTRAKYFIRDEFRLIST
 ASGDGRHYCYPHFTCAVDTENIRRVFNDCRDI IQRMHLRQYELL

SGP**TRRRLEQKLI**SEEDLAANDILDYKDDDDKV

Restriction Sites:

Sgfl-NotI

| | |
|--------------------------|---|
| Protein Pathways: | Calcium signaling pathway, Dilated cardiomyopathy, Gap junction, GnRH signaling pathway, Long-term depression, Melanogenesis, Taste transduction, Vascular smooth muscle contraction, Vibrio cholerae infection |
| MW: | 43.3 kDa |
| Gene Summary: | <p>This locus has a highly complex imprinted expression pattern. It gives rise to maternally, paternally, and biallelically expressed transcripts that are derived from four alternative promoters and 5' exons. Some transcripts contain a differentially methylated region (DMR) at their 5' exons, and this DMR is commonly found in imprinted genes and correlates with transcript expression. An antisense transcript is produced from an overlapping locus on the opposite strand. One of the transcripts produced from this locus, and the antisense transcript, are paternally expressed noncoding RNAs, and may regulate imprinting in this region. In addition, one of the transcripts contains a second overlapping ORF, which encodes a structurally unrelated protein - Alex. Alternative splicing of downstream exons is also observed, which results in different forms of the stimulatory G-protein alpha subunit, a key element of the classical signal transduction pathway linking receptor-ligand interactions with the activation of adenylyl cyclase and a variety of cellular responses. Multiple transcript variants encoding different isoforms have been found for this gene. Mutations in this gene result in pseudohypoparathyroidism type 1a, pseudohypoparathyroidism type 1b, Albright hereditary osteodystrophy, pseudopseudohypoparathyroidism, McCune-Albright syndrome, progressive osseous heteroplasia, polyostotic fibrous dysplasia of bone, and some pituitary tumors. [provided by RefSeq, Aug 2012]</p> |