

Product datasheet for RC402278

OriGene Technologies, Inc.

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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: R231H

Affected Codon#: 231

Affected NT#: 692

Nucleotide Mutation: GNAS Mutant (R231H), Myc-DDK-tagged ORF clone of Homo sapiens GNAS complex locus

(GNAS), transcript variant 1 as transfection-ready DNA

Effect: Pseudohypoprhyroidism 1

Symbol: GNAS

Synonyms: AHO; C20orf45; GNAS1; GPSA; GSA; GSP; NESP; PITA3; POH; SCG6; SgVI

E. coli Selection: Kanamycin (25 ug/mL)

Mammalian Cell Neomycin

Selection:

Vector: pCMV6-Entry (PS100001)

Tag: Myc-DDK
ACCN: NM 000516

ORF Size: 1182 bp
Restriction Sites: Sgfl-Notl

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ORF Nucleotide Sequence:

>RC402278 representing NM_000516
Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCCGCCGCGATCGCC

ATGGGCTGCCTCGGGAACAGTAAGACCGAGGACCAGCGCAACGAGGAGAAGGCGCAGCGTGAGGCCAACA AAAAGATCGAGAAGCAGCTGCAGAAGGACAAGCAGGTCTACCGGGCCACGCACCGCCTGCTGCTGCTGGG TGCTGGAGAATCTGGTAAAAGCACCATTGTGAAGCAGATGAGGATCCTGCATGTTAATGGGTTTAATGGA GAGGGCGGCGAAGAGGACCCGCAGGCTGCAAGGAGCAACAGCGATGGTGAGAAGGCAACCAAAGTGCAGG ACATCAAAAACAACCTGAAAGAGGCGATTGAAACCATTGTGGCCGCCATGAGCAACCTGGTGCCCCCCGT GGAGCTGGCCAACCCCGAGAACCAGTTCAGAGTGGACTACATCCTGAGTGTGATGAACGTGCCTGACTTT AACGCTCCAACGAGTACCAGCTGATTGACTGTGCCCAGTACTTCCTGGACAAGATCGACGTGATCAAGCA GGCTGACTATGTGCCGAGCGATCAGGACCTGCTTCGCTGCCGTGTCCTGACTTCTGGAATCTTTGAGACC GGATCCAGTGCTTCAACGATGTGACTGCCATCATCTTCGTGGTGGCCAGCAGCAGCAGCTACAACATGGTCAT CCGGGAGGACAACCAGACCAACCGCCTGCAGGAGGCTCTGAACCTCTTCAAGAGCATCTGGAACAACAGA GGAAATCGAAGATTGAGGACTACTTTCCAGAATTTGCTCGCTACACTACTCCTGAGGATGCTACTCCCGA GCCCGGAGAGGACCCACGCGTGACCCGGGCCAAGTACTTCATTCGAGATGAGTTTCTGAGGATCAGCACT GCCAGTGGAGATGGGCGTCACTACTGCTACCCTCATTTCACCTGCGCTGTGGACACTGAGAACATCCGCC GTGTGTTCAACGACTGCCGTGACATCATTCAGCGCATGCACCTTCGTCAGTACGAGCTGCTC

AGCGGACCGACGCGTACGCGGCCGCTCGAGCAGAAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCC
TGGATTACAAGGATGACGACGA TAAGGTTTAA

Protein Sequence:

>RC402278 representing NM_000516 Red=Cloning site Green=Tags(s)

MGCLGNSKTEDQRNEEKAQREANKKIEKQLQKDKQVYRATHRLLLLGAGESGKSTIVKQMRILHVNGFNG EGGEEDPQAARSNSDGEKATKVQDIKNNLKEAIETIVAAMSNLVPPVELANPENQFRVDYILSVMNVPDF DFPPEFYEHAKALWEDEGVRACYERSNEYQLIDCAQYFLDKIDVIKQADYVPSDQDLLRCRVLTSGIFET KFQVDKVNFHMFDVGGQRDEHRKWIQCFNDVTAIIFVVASSSYNMVIREDNQTNRLQEALNLFKSIWNNR WLRTISVILFLNKQDLLAEKVLAGKSKIEDYFPEFARYTTPEDATPEPGEDPRVTRAKYFIRDEFLRIST ASGDGRHYCYPHFTCAVDTENIRRVFNDCRDIIQRMHLRQYELL

SGPTRTRRLEQKLISEEDLAANDILDYKDDDDK**V**

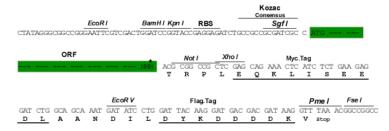
Restriction Sites:

Sgfl-Notl



Cloning Scheme:





^{*} The last codon before the Stop codon of the ORF

OTI Disclaimer:

Due to the inherent nature of this plasmid, standard methods to replicate additional amounts of DNA in E. coli are highly likely to result in mutations and/or rearrangements. Therefore, OriGene does not guarantee the capability to replicate this plasmid DNA. Additional amounts of DNA can be purchased from OriGene with batch-specific, full-sequence verification at a reduced cost. Please contact our customer care team at custsupport@origene.com or by calling 301.340.3188 option 3 for pricing and delivery.

The molecular sequence of this clone aligns with the gene accession number as a point of reference only. However, individual transcript sequences of the same gene can differ through naturally occurring variations (e.g. polymorphisms), each with its own valid existence. This clone is substantially in agreement with the reference, but a complete review of all prevailing variants is recommended prior to use. More info

OTI Annotation:

This clone was engineered to express the complete ORF with an expression tag. Expression varies depending on the nature of the gene.

Components:

The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).

RefSeq: NP 000507 RefSeq Size: 1182 bp RefSeq ORF: 1185 bp Locus ID: 2778 Cytogenetics: 20q13.32

Domains: G-alpha

Protein Families: Druggable Genome, Secreted Protein



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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: 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 reponses. 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 osseus heteroplasia, polyostotic fibrous dysplasia of bone, and some pituitary

tumors. [provided by RefSeq, Aug 2012]