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Product datasheet for RC402256

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:	Q12X
Affected Codon#:	12
Affected NT#:	34
Nucleotide Mutation:	GNAS Mutant (Q12X), Myc-DDK-tagged ORF clone of Homo sapiens GNAS complex locus (GNAS), transcript variant 1 as transfection-ready DNA
Effect:	Proressive osseous heeroplsi
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:	33 bp
Restriction Sites:	SgfI-NotI
ORF Nucleotide Sequence:	>RC402256 representing NM_000516 Red=Cloning site Blue=ORF Green=Tags(s) TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC GCCGCGATCGCC ATGGGCTGCCTCGGGAACAGTAAGACCGAGGAC AGCGGACCGACGCGTACGCGGCCGCTCGAGCAGAAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCC TGGATTACAAGGATGACGACGA TAAGGTTTAA

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Protein Sequence:

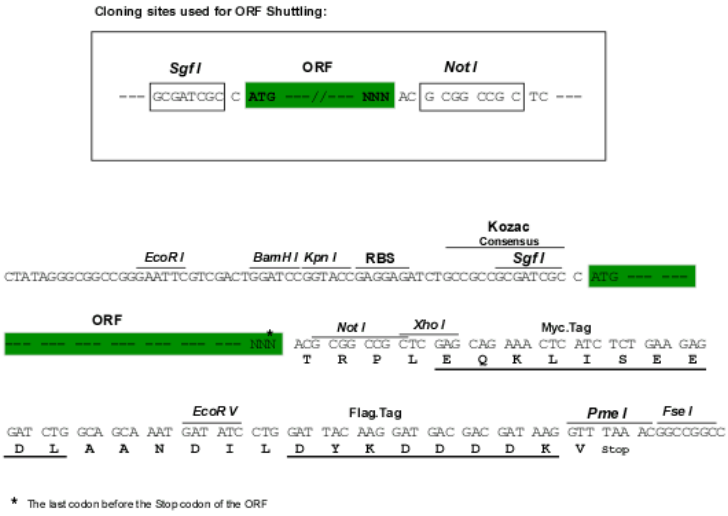
>RC402256 representing NM_000516
Red=Cloning site Green=Tags(s)

MGCLGNSKTED

SGPTRTRRLEQKLISEEDLAANDILDYKDDDDKV

Restriction Sites: SgfI-NotI

Cloning Scheme:



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: 33 bp

RefSeq ORF: 1185 bp

Locus ID:	2778
Cytogenetics:	20q13.32
Domains:	G-alpha
Protein Families:	Druggable Genome, Secreted Protein
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:	1.2 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]