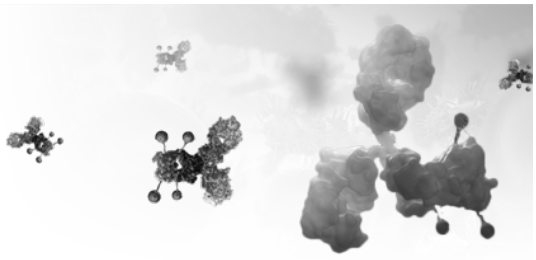

# Human GNAS blocking peptide (CDBP1383)

This product is for research use only and is not intended for diagnostic use.

## PRODUCT INFORMATION

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|---|---|
| **Product Overview** | Blocking/Immunizing peptide for anti-GNAS antibody |
| **Antigen Description** | 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. |
| **Species** | Human |
| **Conjugate** | Unconjugated |
| **Applications** | Apuri, BL, ELISA |
| **Format** | Lyophilized powder |
| **Size** | 100 µg |
| **Preservative** | None |
| **Storage** | Shipped at ambient temperature, store at -20°C. |

45-1 Ramsey Road, Shirley, NY 11967, USA
Email: info@creative-diagnostics.com
Tel: 1-631-624-4882 Fax: 1-631-938-8221

# GENE INFORMATION

| | |
|---|---|
| **Gene Name** | GNAS GNAS complex locus [ Homo sapiens ] |
| **Official Symbol** | GNAS |
| **Synonyms** | GNAS; GNAS complex locus; GNAS1, guanine nucleotide binding protein (G protein), alpha stimulating activity polypeptide 1; protein ALEX; GNASXL; GPSA; NESP; NESP55; SCG6; secretogranin VI; extra large alphas protein; neuroendocrine secretory protein; guanine nucleotide regulatory protein; alternative gene product encoded by XL-exon; adenylate cyclase-stimulating G alpha protein; guanine nucleotide-binding protein G(s) subunit alpha isoforms XLas; guanine nucleotide binding protein (G protein), alpha stimulating activity polypeptide 1; AHO; GSA; GSP; POH; GNAS1; PHP1A; PHP1B; PHP1C; C20orf45; MGC33735; |
| **Entrez Gene ID** | 2778 |
| **mRNA Refseq** | NM_000516 |
| **Protein Refseq** | NP_000507 |
| **UniProt ID** | O95467 |
| **Chromosome Location** | 20q13.2-q13.3 |
| **Pathway** | Amoebiasis, organism-specific biosystem; Amoebiasis, conserved biosystem; Amphetamine addiction, organism-specific biosystem; Amphetamine addiction, conserved biosystem; Aquaporin-mediated transport, organism-specific biosystem; Bile secretion, organism-specific biosystem; Bile secretion, conserved biosystem; |
| **Function** | G-protein beta/gamma-subunit complex binding; GTP binding; GTPase activity; GTPase activity; adenylate cyclase activity; guanyl nucleotide binding; guanyl-nucleotide exchange factor activity; ionotropic glutamate receptor binding; metal ion binding; molec |

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