

Recombinant Human GNAS

Catalog No: #GP10425



Package Size: #GP10425-1 100ug

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Description

Product Name	Recombinant Human GNAS
Brief Description	Recombinant Protein
Immunogen Description	Fusion protein corresponding to C terminal 200 amino acids of human GNAS complex locus
Target Name	GNAS complex locus
Other Names	AHO, GSA, GSP, POH, GPSA, NESP, GNAS1, PHP1A, PHP1B, PHP1C, C20orf45
Accession No.	Swissprot:P63092Gene Accession:BC002722
Uniprot	P84996
GeneID	2778;
Storage	-20~-80°C, pH 7.6 PBS

Background

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.?

References

Note: For in vitro research use only, not for diagnostic or therapeutic use. This product is not a medical device.

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