

GNAS

Reactivity: Human Mouse Rat

Tested applications: WB

Recommended Dilution: WB 1:500 - 1:2000

Calculated MW: 46kDa

Observed MW: Refer to Figures

Immunogen:

Recombinant protein of human GNAS

Storage Buffer:

Store at -20. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

Synonym:

AHO; GSA; GSP; POH; GPSA; NESP; GNAS1; PHP1A; PHP1B; PHP1C; C20orf45

Catalog #: A2732

Antibody Type:

Polyclonal Antibody

Species: Rabbit

Gene ID: 2778

Isotype: IgG

Swiss Prot: P63092

Purity: Affinity purification

For research use only.

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. [provided by RefSeq, Aug 2012]

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