SANTA CRUZ BIOTECHNOLOGY, INC.

NBEAL2 (N-14): sc-243582

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The Power to Question

BACKGROUND

NBEAL2 (neurobeachin-like protein 2), also known as GPS or BDPLT4, is a 2,754 amino acid protein that belongs to the WD repeat neurobeachin family. Existing as four alternatively spliced isoforms, NBEAL2 contains one BEACH domain and five WD repeats. NBEAL2 is thought to play a role in α-granule biogenesis in megakaryocytes and is encoded by a gene that maps to human chromosome 3p21.31. Mutations of the NBEAL2 gene are the cause of gray platelet syndrome (GPS), an autosomal recessive platelet disorder in which large platelets that lack α-granules are observed. Patients with GPS may exhibit moderate to severe bleeding. Chromosome 3 houses over 1,100 genes, in-cluding a chemokine receptor (CKR) gene cluster and a variety of human cancer-related gene loci. Marfan Syndrome, osteogenesis imperfecta and Charcot-Marie-Tooth disease are a few of the numerous genetic diseases associated with chromosome 3.

REFERENCES

1. De Jonghe, P., et al. 1997. Mutilating neuropathic ulcerations in a chromosome 3q13-q22 linked Charcot-Marie-Tooth disease type 2B family. J. Neurol. Neurosurg. Psychiatr. 62: 570-573.
2. Maho, A., et al. 1999. Mapping of the CCXCR1, CX3CR1, CCBP2 and CCR9 genes to the CCR cluster within the 3p21.3 region of the human genome. Cytogenet. Cell Genet. 87: 265-268.
3. Robinson, P.N., et al. 2000. The molecular genetics of Marfan syndrome and related microfibrillopathies. J. Med. Genet. 37: 9-25.
4. Gunay-Aygun, M., et al. 2010. Gray platelet syndrome: natural history of a large patient cohort and locus assignment to chromosome 3p. Blood 116: 4990-5001.
5. Gunay-Aygun, M., et al. 2011. NBEAL2 is mutated in gray platelet syndrome and is required for biogenesis of platelet α-granules. Nat. Genet. 43: 732-734.
6. Albers, C.A., et al. 2011. Exome sequencing identifies NBEAL2 as the causative gene for gray platelet syndrome. Nat. Genet. 43: 735-737.
7. Kahr, W.H., et al. 2011. Mutations in NBEAL2, encoding a BEACH protein, cause gray platelet syndrome. Nat. Genet. 43: 738-740.

CHROMOSOMAL LOCATION

Genetic locus: NBEAL2 (human) mapping to 3p21.31; Nbeal2 (mouse) mapping to 9 F2.

SOURCE

NBEAL2 (N-14) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of NBEAL2 of human origin.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-243582 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

NBEAL2 (N-14) is recommended for detection of NBEAL2 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000); non cross-reactive with NBEAL1.

Suitable for use as control antibody for NBEAL2 siRNA (m): sc-149847, NBEAL2 shRNA Plasmid (m): sc-149847-SH and NBEAL2 shRNA (m) Lentiviral Particles: sc-149847-V.

Molecular Weight of NBEAL2 isoforms: 302/283/112/67 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

For research use only, not for use in diagnostic procedures.

PROTOCOLS

See our web site at www.scbt.com or our catalog for detailed protocols and support products.