SANTA CRUZ BIOTECHNOLOGY, INC.

RPGRIP1L (N-15): sc-165400

BACKGROUND

RPGRIP1L (RPGRIP1-like), also known as protein fantom, NPHP8 (nephrocystin 8), MKS5, CORS3, JBTS7 or FTM, is a 1,315 amino acid protein that belongs to the RPGRIP1 family and is thought to function in programmed cell death, craniofacial development and formation of the left-right axis. Existing as two alternatively spliced isoforms that localize to the cytoplasm, cytoskeleton, centrosome and cilium basal body, RPGRIP1L interacts with nephrocystin-4 and is moderately expressed in brain, retina and kidney. Containing two C2 domains, RPGRIP1L is encoded by a gene that maps to human chromosome 16q12.2. Defects in the gene encoding RPGRIP1L are the cause of Joubert syndrome type 7 (JBTS7), COACH syndrome (COACHS) and Meckel syndrome type 5 (MKS5).

REFERENCES

1. Wolf, M.T., et al. 2007. Mutational analysis of the RPGRIP1L gene in patients with Joubert syndrome and nephronophthisis. Kidney Int. 72: 1520-1526.
2. Delous, M., et al. 2007. The ciliary gene RPGRIP1L is mutated in cerebello-oculo-renal syndrome (Joubert syndrome type B) and Meckel syndrome. Nat. Genet. 39: 875-881.
3. Arts, H.H., et al. 2007. Mutations in the gene encoding the basal body protein RPGRIP1L, a nephrocystin-4 interactor, cause Joubert syndrome. Nat. Genet. 39: 882-888.
4. Brancati, F., et al. 2008. RPGRIP1L mutations are mainly associated with the cerebello-renal phenotype of Joubert syndrome-related disorders. Clin. Genet. 74: 164-170.
5. Khanna, H., et al. 2009. A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nat. Genet. 41: 739-745.
6. Doherty, D., et al. 2010. Mutations in 3 genes (MKS3, CC2D2A and RPGRIP1L) cause COACH syndrome (Joubert syndrome with congenital hepatic fibrosis). J. Med. Genet. 47: 8-21.
7. Online Mendelian Inheritance in Man, OMIM™. 2010. Johns Hopkins University, Baltimore, MD. MIM Number: 610937. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/

CHROMOSOMAL LOCATION

Genetic locus: RPGRIP1L (human) mapping to 16q12.2; Rpgrip1l (mouse) mapping to 8 C5.

SOURCE

RPGRIP1L (N-15) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the N-terminus of RPGRIP1L 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-165400 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

RPGRIP1L (N-15) is recommended for detection of RPGRIP1L 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 RPGRIP1.

RPGRIP1L (N-15) is also recommended for detection of RPGRIP1L in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for RPGRIP1L siRNA (h): sc-93387, RPGRIP1L siRNA (m): sc-153102, RPGRIP1L shRNA Plasmid (h): sc-93387-SH, RPGRIP1L shRNA Plasmid (m): sc-153102-SH, RPGRIP1L shRNA (h) Lentiviral Particles: sc-93387-V and RPGRIP1L shRNA (m) Lentiviral Particles: sc-153102-V.

Molecular Weight of RPGRIP1L isoforms: 151/143 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.