

# RPGRIP1L (E-14): sc-165399

## BACKGROUND

RPGRIP1L (RPGRIP1-like), also known as protein phantom, 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<sup>™</sup>. 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 (E-14) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region 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-165399 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

RPGRIP1L (E-14) 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), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], 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.

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.

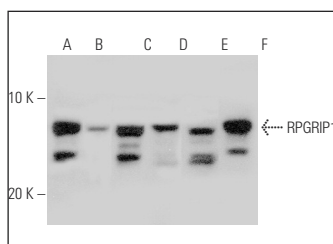
Molecular Weight of RPGRIP1L isoforms: 151/143 kDa.

Positive Controls: HEK293 whole cell lysate: sc-45136, NTERA-2 cl.D1 whole cell lysate: sc-364181 or HeLa whole cell lysate: sc-2200.

## 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<sup>™</sup> compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker<sup>™</sup> Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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<sup>™</sup> Mounting Medium: sc-24941.

## DATA



RPGRIP1L (E-14): sc-165399. Western blot analysis of RPGRIP1L expression in NTERA-2 cl.D1 (A), HEK293 (B), HeLa (C) and Jurkat (D) whole cell lysates and human brain (E) and human kidney (F) tissue extracts.

## 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.