

RPGRIP1 (m): 293T Lysate: sc-123274

BACKGROUND

RPGRIP1 (retinitis pigmentosa GTPase regulator interacting protein 1), also known as LCA6, RGI1, RGRIP, CORD13, RPGRIP or RPGRIP1d, is a 1,286 amino acid protein that belongs to the RPGRIP1 family and localizes to the cilium. Expressed in retina, RPGRIP1 colocalizes with SEC16S in the outer segment of rod photoreceptors and cone outer segments. RPGRIP1 forms homodimers and elongated homopolymers, and exists six alternatively spliced isoforms. RPGRIP1 is required for SEC16S function and is essential for normal disk morphogenesis. Mutations in the gene encoding RPGRIP1 are the cause of Leber congenital amaurosis type 6 (LCA6) and cone-rod dystrophy type 13 (CORD13). LCA is considered the most common genetic cause of congenital visual impairment in infants and children. CORD13 is an inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination and initial loss of cone photoreceptors followed by rod degeneration.

REFERENCES

- Boylan, J.P., et al. 2000. Identification of a novel protein interacting with RPGR. *Hum. Mol. Genet.* 9: 2085-2093.
- Roepman, R., et al. 2000. The retinitis pigmentosa GTPase regulator (RPGR) interacts with novel transport-like proteins in the outer segments of rod photoreceptors. *Hum. Mol. Genet.* 9: 2095-2105.
- Gerber, S., et al. 2001. Complete exon-intron structure of the RPGR-interacting protein (RPGRIP1) gene allows the identification of mutations underlying Leber congenital amaurosis. *Eur. J. Hum. Genet.* 9: 561-571.
- Mavlyutov, T.A., et al. 2002. Species-specific subcellular localization of RPGR and RPGRIP1 isoforms: implications for the phenotypic variability of congenital retinopathies among species. *Hum. Mol. Genet.* 11: 1899-1907.
- Hameed, A., et al. 2003. Evidence of RPGRIP1 gene mutations associated with recessive cone-rod dystrophy. *J. Med. Genet.* 40: 616-619.
- Pawlyk, B.S., et al. 2010. Replacement gene therapy with a human RPGRIP1 sequence slows photoreceptor degeneration in a murine model of Leber congenital amaurosis. *Hum. Gene Ther.* 21: 993-1004.
- Fernández-Martínez, L., et al. 2011. Evidence for RPGRIP1 gene as risk factor for primary open angle glaucoma. *Eur. J. Hum. Genet.* 19: 445-451.
- Kuznetsova, T., et al. 2011. Structural organization and expression pattern of the canine RPGRIP1 isoforms in retinal tissue. *Invest. Ophthalmol. Vis. Sci.* 52: 2989-2998.
- Busse, C., et al. 2011. Ophthalmic and cone derived electrodiagnostic findings in outbred Miniature Long-haired Dachshunds homozygous for a RPGRIP1 mutation. *Vet. Ophthalmol.* 14: 146-152.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

PROTOCOLS

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

CHROMOSOMAL LOCATION

Genetic locus: Rpgrip1 (mouse) mapping to 14 C2.

PRODUCT

RPGRIP1 (m): 293T Lysate represents a lysate of mouse RPGRIP1 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

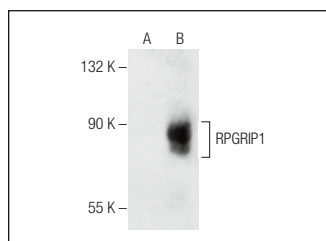
RPGRIP1 (m): 293T Lysate is suitable as a Western Blotting positive control. Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

RPGRIP1 (C-9): sc-390341 is recommended as a positive control antibody for Western Blot analysis of enhanced mouse RPGRIP1 expression in RPGRIP1 transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

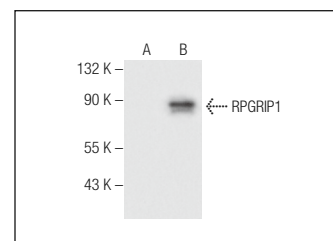
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended:
1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



RPGRIP1 (C-9): sc-390341. Western blot analysis of RPGRIP1 expression in non-transfected: sc-117752 (A) and mouse RPGRIP1 transfected: sc-123274 (B) 293T whole cell lysates.



RPGRIP1 (B-3): sc-390331. Western blot analysis of RPGRIP1 expression in non-transfected: sc-117752 (A) and mouse RPGRIP1 transfected: sc-123274 (B) 293T whole cell lysates.

RESEARCH USE

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